U.S. DEPARTMENT OF HEALTH AND HUMAN SERVICES
ADVISORY COMMITTEE ON HERITABLE DISORDERS AND GENETIC DISEASES IN NEWBORNS AND CHILDREN
Third Meeting
Thursday, January 13, 2005
Rotunda Room, 8th Floor
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1300 Pennsylvania Avenue, N.W.
Washington, D.C.
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Bradford L. Therrell, Ph.D.
Director
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Stephen C. Groft, Pharm.D.
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Discussion

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Michele A. Lloyd-Puryear, M.D., Ph.D.
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Judith M. Tuerck, R.N., M.S.
Assistant Professor
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David Ross, Sc.D.
Director
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Peter C. van Dyck, M.D., M.P.H.
Associate Administrator
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Peter C. van Dyck, M.D., M.P.H.

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P R O C E E D I N G S (9:09 a.m.)

DR. HOWELL: Ladies and gentlemen, could I ask that you have a seat so we can get started?
Let me introduce Dr. Dennis Williams, who will bring greetings from HHS and give us some direction for the day.

DR. WILLIAMS: Thank you, Mr. Chairman, and welcome to the committee. It's really my pleasure to welcome the members of the Advisory Committee on Heritable Disorders and Genetic Diseases in Newborns and Children. Secretary Thompson and HRSA Administrator Duke also send their greetings to you.
As many of you know, Secretary Thompson will be leaving HHS soon. President Bush has asked former Utah Governor Mike Leavitt to take over as Secretary once his confirmation hearings are over. At this time I want to take the opportunity to say how fortunate we have been to have Secretary Thompson's fine leadership on issues that relate to the welfare of the nation's mothers and children. For all of us at HHS, it's been a pleasure to work for him, and in the months ahead we can honor his service best by keeping up the good work.

Today I also want to thank Dr. Rodney Howell of the University of Miami School of Medicine for his willingness to give so much of his time and energy in his service as chair of this committee. I want to thank Dr. Peter van Dyck, HRSA Associate Administrator for Maternal and Child Health, for his leadership as well. Finally, I want to welcome the newest member of the committee, Dr. Joseph Telfair, who is representing the Secretary's Advisory Committee on Genetics, Health, and Society. Welcome.
To all of you I want to emphasize once again how much we value the work you are doing. You provide critical advice on the necessary steps states can take to assure that all Americans' children receive quality health care. Today you'll be evaluating many critical issues surrounding newborn screening programs, opportunities for research, information systems for evaluating the long-term health outcomes of the infants identified, surveillance systems for evaluating newborn screening programs, and the Regional Collaboratives established this past year by HRSA's Maternal and Child Health Bureau.

At previous meetings, I know you discussed the financing of newborn screening programs and how states are using new technologies, such as tandem mass spectrometry. I urge you all to weigh in on these issues and set priorities for the next year.

The Regional Collaboratives funded by the Maternal and Child Health Bureau across the country have been established to provide a network of genetic service providers to shift services into local communities. This network of seven Regional Collaboratives in a National Coordinating Center is a necessary part of the entire newborn screening system. Dr. Michele Puryear will give you an update later on this new crucial activity.

In closing, I can tell you that we at HRSA look forward to your recommendations on how we can best improve newborn screening services nationwide. We're very confident that your knowledge and expertise will help us recommend guidelines to Secretary Thompson's successor that will greatly benefit children and their families across the United States. Once again, we thank you for your willingness to participate and wish you good luck in your meeting.

DR. HOWELL: Thank you very much, Dr. Williams. Again, on behalf of the committee, let me thank you and the leadership at the Maternal and Child Health Bureau and at HRSA for the support that you've provided to this committee.

I again express my appreciation at the beginning of this meeting for the hard work that the committee members have continued to provide in this area.

The first item of business that we have to do is to approve the minutes of the last meeting, which is in your agenda book. Can we hear some comments and recommendations about the minutes?

(No response.)

DR. HOWELL: Is there a motion to approve the minutes of the last meeting as they're presented?

PARTICIPANT: So moved.

DR. HOWELL: Second?

DR. COGGINS: Second.

DR. HOWELL: Peter.
All in favor of that?

(Chorus of ayes.)

DR. HOWELL: That's a uniform thing.
I think that you have in your book the agenda, which is an extensive and a busy one. I think we're going to focus a lot during this time on some of the early issues that we come upon as far as the infrastructure for newborn screening. We have focused considerable time up until now about decision-making about what might be on the panel. We're going to come back to that also later in the meeting. But also, you will see that we have a series of important presentations, one of which has been mentioned on the Regional Genetics and Newborn Screening Collaboratives that Dr. Puryear will discuss. But we're going to hear a lot about infrastructure and data management programs that I think will be important for us to hear about and weigh in on.

Again, let me formally introduce Dr. Joseph Telfair, who is newly appointed to the Secretary's Advisory Committee on Genetics, Health, and Society.
Dr. Telfair, we welcome you to this committee and look forward to your participation. Would you like to make any comments?

DR. TELFAIR: Just to say thank you for so graciously extending your greetings, and I hope I can

contribute the best I can.

DR. HOWELL: I think your experience in some of the very important areas of newborn screening will be invaluable as we look at some of those issues.

On the agenda it talks about public comment, and we're going to have general public comment later, but are there any comments at this point that anybody would like to make before we start into the heart of the agenda today?
Dr. Howse?

DR. HOWSE: Thank you, Dr. Howell.
I was not able to attend the September meeting of the committee as I was traveling in China, but I did send an extensive letter on behalf of the March of Dimes with respect to the disposition of the report.

I have two comments to make. One is I trust that we will find time during the course of these two days to discuss the disposition of the report. Our organization feels it's quite important -- I know a number of others do as well -- that the report get into the public domain for public comment in its present form. So that would be the first issue, and that I think is quite important to move the program ahead.

Then secondly, that we pick up what was mentioned in the minutes of our first meeting and reflected again in September, that we really give some serious thought to the subcommittee structure for this committee and really identify the headers that most centrally address where we feel the program needs to go so we can do work in-between times and come to this meeting with recommendations and ideas and action.

So I'd just ask that those two items at some point in the two days be covered.

DR. HOWELL: Thank you very much. I think that, if you will note on the agenda, we're going to have an extensive discussion about the report and its disposition. Dr. van Dyck is going to lead that discussion because there's been tremendous interest in the status of the report that we heard about and its disposition, and I think that before we leave we should have a very clear understanding and idea of exactly what's happening with that and so forth, because I think that we share your interest in that. Secondly, again, a major item is the setting committee priorities, which again will be a formal part of the agenda, and we should come back to that because the issues we're going to be dealing with, and there are an enormous number of major areas of interest that this committee should deal with, and I think the key thing will be to identify a group that is dealable with, for want of a better word, so that we can move ahead on certain key issues and so forth. But I would hope that we would leave with both of those under considerable solution.

Thank you very much.

Any further comments about the agenda and what we would hope to accomplish while we're here?

(No response.)

DR. HOWELL: If not, we're going to start off this morning and hear from Dr. Brad Therrell, who is Director of the National Newborn Screening and Genetics Resource Center. As everybody in this room and particularly on this panel knows, the status of newborn screening changes daily, and Brad and his group keep a very detailed documentation of what's happening. So we'll hear from Brad about newborn screening activities.

DR. THERRELL: Thank you, Dr. Howell.

Dr. Williams, committee, it's my pleasure to be here again to visit with you and bring you a little bit more information about what's been happening since the last time you met in the world of newborn screening. I'd like to begin by acknowledging the help of my able-bodied assistant, Donna Williams, who is here today in helping me prepare this information. She's really the person who keeps contact with the states on the telephone and on the computer.

I'd also like to acknowledge the help of the Association of Public Health Laboratorians, which has a listserv that reaches out to the 51 programs in the states, as opposed to the listserv which we have which reaches out to 500 people around the world. So when we want to keep it small, we use APHL's, and we wanted to keep it small this time.

I'd also like to draw your attention to Tab 5, which is where my slide presentation is. At the end of that tab are two tables about what states are doing in newborn screening, and you may want to look at those as I go through the presentation.

What I thought we would do just to bring you up to date is to go through what some of the states have told us, and I hope this is comprehensive, but it's only as comprehensive as people voluntarily report information to us, and that's an issue that was addressed in the ACMG report, and I hope you'll discuss that a little bit more. There's no mandated way that programs report. It's all voluntary. So we get information from all sorts of sources, and we validate it all the time with the states. So we may not have the latest information that somebody in a particular state in this room knows about. We'd be happy to take that after the meeting.

So what I've done is I have a series of slides where I put two or three states to a slide with some of the information that they gave us in response to our request: "What have you been doing new in the last few months?" So from Alabama, we have information that they have now implemented biotinidase deficiency and six additional MS/MS disorders during the last quarter of the year.

In California, there's been big news in California. They were mandated last year to go ahead and add tandem mass spectrometry, and they're beginning to do that. It takes a while to set this program up. So the first step was a fee increase, which did go into effect on January 1st, and their fee raised from $60 to $78. Talking to Dr. Cunningham, this is normally done six months before they implement the program. So we would expect in six months to see this program being implemented. They will add 32 MS/MS disorders, in their minds raising the total to 87. This is an issue that we'll also want to talk about at this meeting, how do you count conditions, how do you name conditions. But the release of information that initially went out was 87 disorders. CF and biotinidase deficiency are not on the list, and they hope to be added later.

Colorado began some MS/MS work last year at the University of Colorado, and they hope that by the summer of this year their legislature will actually consider the issues of tandem mass spectrometry and add those. They have asked our center to provide them with a review of their program in the next few months, which is, as you'll recall, one of the things that our center does, responds to requests from state health departments to go in and analyze their program and answer their questions and issues and give them information that they can use to further improve their programs.

In Connecticut, early in 2004 their law changed so that they were supposed to increase from five disorders to over 30 disorders. So May 1st -- they phased this in. On May 1st they added three conditions and increased the fee to $28, but they dropped their second mandatory sample at the same time. By October 1st, they had added seven more conditions. On November 1st, they added seven more. On January 1st, they added 13 more. So they've done everything they were supposed to do. CF is not included in their mandate to expand. Now, CF is done voluntarily in a project at the University of Connecticut, and that continues, and that reaches about 80 percent of the patients.

Florida also big news. They have a mandate to increase their program from five to over 30 disorders, and they anticipate doing that by February 1st, and they did include CF and biotinidase deficiency.

Georgia finally got going. They have been mandated to do this for a while, but on January 1st they did begin screening for MCAD deficiency.

Indiana, in 2004 they increased their fee to $62.50, which included a $32.50 laboratory surcharge, and they're also working on continuing to expand their program.

Iowa had a co-change in 2004 which changed the name of their center that does newborn screening from the Birth Defects Institute to the Center for Congenital and Inherited Disorders. In 2005 they're beginning a CF pilot project. They're going to be moving to a new facility that has refrigerated storage for their blood spots.

Louisiana started an MS/MS pilot, November the 1st added five MS/MS disorders, and on the first day they picked up a citrullinemia. Within the first month, they picked up two MCADs. So right now this is still in the pilot phase. By July 1st they anticipated a real change that will make this effective in law, and their fee will increase at the time they anticipate to at least $40.

Michigan added homocystinuria, citrullinemia and ASA October the 1st, and their fee also increased, and they also changed their metabolic center from a contract with University of Michigan to a contract with Wayne State.

Minnesota began a pilot for biotinidase deficiency on September the 9th and officially added biotinidase on January 3rd of this year.
Missouri, the information they sent us was that they will increase from five to about 25 early in this year, and we don't have further details.

Nebraska has had some interesting lawsuits which I thought would be of interest to this committee. Nebraska has a law which does not allow for dissent. They're one of about five states that does not allow dissent from newborn screening for any reason. So that's been challenged a couple of times in the court previously, and always the state had won those cases. Recently the same thing happened. There was a suit, the state won, but now that one's been appealed and it's supposed to go to the Supreme Court in Nebraska.

Also, there's another lawsuit that's been instigated there about religious discrimination. Their law and rules combined allow if a baby is born in an ICU, that the ICU has to send in a sample, but they have up to seven days to do it, which is consistent with AAP recommendations for babies who are in stress. So this group has filed suit and actually, I'm told, has gotten an injunction which allows them to sort of opt out of the seven-day period for religious reasons. So now they can claim that this is discriminatory because it allows the ICU to wait seven days, but it doesn't allow anybody else, and the judge has right now enjoined the state from mandating that anybody else has to do it too. That yet has to be resolved. It's gotten to be pretty messy, I'm told.

New Hampshire is in the process of adding four conditions, so that they'll now have a total of 10. The big news here is that they've added universal screening for sickle cell disease, which they didn't have previously.

New York, we have several quotes from the Governor's office. In 2004 they had a press release that said we will increase to 31 by the end of 2004, and then we'll have additional expansion by spring to 44 conditions. If you look at the list of 44 conditions, you'll find that there's one that's counted twice, so it's really 43. In the State of the State address that was just given, this is a quote at the bottom: "This year, one of the new tests will be Krabbe disease." I've tried to get confirmation that that's actually going to be on the panel from New York. I think it's only in the pilot phase, but I haven't gotten confirmation back.

North Carolina began biotinidase deficiency in November, and they're exploring the possibility of adding CF.

Ohio anticipates the addition of CF and has begun or is beginning the CF validation process.

Oklahoma is trying to implement CAH, CF and MCAD. We thought it was going, but we talked to them the other day and it's not yet going. There's some discussion still ongoing about final implementation steps. They expect an increase in the fee, but that has also not been totally worked out at this point.

Oregon convened a task force in 2004 to study and make recommendations on CF, and I heard last night that this committee may have now met and voted to do that, but I don't have confirmation of that from the program. They have purchased a new laboratory building. They'll be combining their public health and environmental quality laboratories together during the coming year, and that will include newborn screening.

Pennsylvania last year added a contract laboratory. Pennsylvania contracts out their laboratory services, and previously they had one laboratory doing that. Last year they added a second laboratory, so now the hospital has the choice of which hospital to contract with in Pennsylvania. They've also had a number of staff retirements and replacements, and that program is beginning to reshape itself. They just had a big stakeholders meeting in Harrisburg.

Rhode Island, we just met with their advisory committee a few weeks ago, and they voted to recommend to the larger genetics advisory committee and to the director of health expansion to screen all the disorders in the core panel being recommended by ACMG through this committee. In 2005, which is now, they're beginning the actions that will make that effective.

South Carolina expanded to 30-plus conditions on November the 1st. They had previously been mandated to do that but hadn't yet implemented everything, and they've increased their fee to $42, effective November the 1st.

Texas still mandates screening for five disorders. The Texas Medical Association, March of Dimes, and the Texas Department of Health, which has a new name, which is actually DHHS, the Department of Health and Human Services, had a stakeholders meeting in December. They invited our review team to come in and do an extensive review of that program at the end of February. March of Dimes is working with the legislature in Texas to enact a start-up law which would provide the funding for start-up for mass spec, but the law at this point would not include CF and biotinidase deficiency.

Virginia, in 2004 there was a joint commission on health care that voted to expand from nine to over 30 conditions, and so this will go to the legislature for approval this year, and a fee increase is expected from $32 to $50.

The State of Washington increased by five disorders in the last quarter of the year, but the fee that they have doesn't currently support the whole program, and this fee is set by the legislature. So they're going back to the legislature, asking them to reconsider the fee this year, and they've begun the process of considering whether CF should be added or not.

I didn't mention hearing screening because there's another resource center which handles most of the hearing screening, but Washington did comment that they're looking at whether or not to change from voluntary to a mandated hearing screening program.

So that's sort of a look at the states, the ones that responded to us. I know that there are others out there. For instance, I know that Maryland has decided also to go ahead with CF screening, but I don't have confirmation of that at this point.

So this is a summation of what I've just gone over. I don't think I need to go through it again. But the thing I'll point out here is that you'll see a number of states did add biotinidase deficiency screening, but a number of states also did not. That's an interesting question as to why that's happening. Mostly states have been expanding into mass spec. You didn't hear very much expansion in terms of CAH, for instance.

So this is a map you've seen before which shows -- in September, when you last had your meeting, the states in the darker color have tandem mass spectrometry in operation. Whether or not they have the machine is another question. Some of them send to the location with a star to get the mass spectrometry done. The states in the green either have a pilot ongoing or they have optional testing available, and the ones in blue have nothing going on.
Now I'll show you the updated map, and you'll see that there were just a couple of changes there. Utah has a pilot going on, and Washington State has added a mass spec to their repertoire.

This is also from the last meeting. This is a map showing you the numbers of different disorders in the states. Again, the ones with the most disorders are the ones that are in purple, and the ones with the least are the lighter colors. I'll show you the changes, and they just were little number changes. I don't know if I can back up on here or not. Let me back up. This is the way it was, this is the way it is, and you see the change is New York went to 30, Connecticut went to greater than 30, and Florida went to greater than 30 mandated. Just because they're mandated doesn't necessarily mean they're doing the tests yet, but most of them are.

I had individual maps last time that I showed you of the states that were actually performing the tests. I'm not going to go back through those, except I will show you for MCAD. This is the map last time you saw where the states in purple have MCAD screening mandated, the states that are cross-hatched have it mandated but haven't yet implemented it, and Colorado was kind of a special case because they haven't yet implemented it and they're fighting about it. So I counted them as 0.5. The ones not screening were the ones in gold, and the ones that have it optional are the ones in lavender.

I'll show you now the new map, which really just changes -- let me back up. Just look over at Louisiana, Florida and Georgia. That's where the main changes are. There it was, and there it is.

Now, in terms of how many disorders are done by the different programs, this is the bar graph where the number not in parentheses tells you the number of programs that are mandated, and the number in parentheses tells you the additional programs that either have it as a pilot or have mandated it but haven't yet started it. Let me show you superimposed on that the new one, and just look at the center, MSUD, biotinidase and homocystinuria. That's where the changes mostly are, right in the center.
So let me back up. This is what it was, and this is what it is now. So you see it's growing a little bit in terms of those disorders in tandem mass spec, and a little bit in terms of biotinidase and cystic fibrosis, but it's staying pretty status quo in terms of CAH, and actually it didn't move as much with biotinidase as it did with some of the others. So this is the way it was, this is the way it is. Okay?
That's pretty much it. I'll be happy to answer questions.

DR. HOWELL: Any questions of Brad?
Piero?

DR. RINALDO: I was under the impression that California also is adding CAH.

DR. THERRELL: Yes. I'm sorry. California is adding CAH. You're right. But the big news there is that they're not adding CF and biotinidase.

DR. BOYLE: You brought up two issues. One was mandated reporting by states, or actually to the federal level is what I think you were thinking, as well as how to count the conditions. Addressing the latter one

first, do you feel like what you've presented to us is counting the conditions the same way across states so we have an accurate picture of that? That's the first question.

Then the second is in terms of mandated reporting. Are most states required to report to their state health department and the issue is more reporting in the aggregate at the federal level, or does that not even occur at the state level?

DR. THERRELL: I'll take your second one first. In terms of the reporting, Title V requires some reporting, but it doesn't necessarily cover all of the individual diseases. So internally within a health department, there is some reporting going on. Sometimes it isn't necessarily validated reporting. So the Title V director may call the newborn screening program or the laboratory and say how many positive PKUs did you get, and the laboratory may say 35. If they ask the question how many confirmed PKU cases did you have, the answer may be two. So sometimes there's some question about the validity of the data.

But beyond that, there's very little reporting that's required, and state newborn screening programs traditionally will, for their own quality control benefit, develop their data and report it, but not always. But there's no mandate to do that. That was my point. And there's no mandate to really report anything other than Title V data, and there's no mandate to report in to anybody what's going on. So it's very difficult to keep up.

Now, we've been pretty successful at that. So to answer your other question, we count disorders sort of the traditional way, and we tried to level the playing field when anybody calls us with new disorders and count that the same way. I know that Dr. van Dyck is very interested in this issue and I'm sure we're going to discuss it more later, but this is quite an issue, and you saw when California -- Dr. Cunningham reported 87 conditions, and other states could do that too if they went back and recounted how they do sickle hemoglobinopathies. I mean, that's the biggest question. How do you count the hemoglobinopathies? Do you count it as one thing or do you count it as 35 or 40 or 250 or whatever?

So George was making a point when he did that, and his point was that there's not any consistency out there, and his point being driven to some extent by the private laboratories who want to get business from the consumers, so they tell the consumers they're doing more and more and more. If a state says 44, they'll say 45. The state also wants to compete, so they'll say 46, and it's a never-ending cycle. So hopefully this committee will address that in some detail.

DR. HOWSE: Brad, the ongoing broil over how to count tests notwithstanding, from your report I think we can see that newborn screening is alive and well in states and that it's a very happening kind of legislative and public health activity that is growing. I wonder if you would comment from your vantage point on the major drivers to the expansion of newborn screening programs, and if you care to do this, just give us a sense of the future and how you think that map is going to look, say, in two to three years.

DR. THERRELL: Well, I think the major driver right now is consumer interest and consumer advocacy, and that really has -- if you look at the history of newborn screening, that's been the driver all along, although it's just getting a lot of publicity in more recent years as we've gotten into more and more expanded technology and expanded knowledge about diseases. So I think the number-one thing is consumer interest.

If you look at those states who haven't yet expanded, you'll find that the consumer interest level in those states hasn't been that high. It's raising and there are consumer advocacy groups around the country that are helping the states where there's not advocacy get moving, but one of the reasons that biotinidase hasn't moved forward, I think, is because there's not really a biotinidase deficiency advocacy group, and it doesn't get rolled into the mass spec issues.

I think that we're going to continue to see expansion. I think that there's a lot of pressure coming from consumers through the press, the press through the legislature, the legislature through the health

departments. So I think you're going to see continued expansion, and I can tell you that our group normally does two or three reviews a year, and right now I've got five waiting, because internally this question comes up, why haven't you expanded, and the programs for the most part have been working hard to do expansion over the years and haven't gotten the support they've needed. So the programs are saying, well, let's have somebody from the outside come in and take a look and validate what we've been doing and tell us what we should be doing differently, especially the big states.

We just did a review of Florida, we've got a review scheduled in Texas, we've got one requested in California. Some of the states that you didn't see reported, we've got one requested in Arizona. So there's a lot of interest in newborn screening, and there's going to continue to be.

DR. BECKER: Brad, could you take a moment to describe for us the web-based reporting system that you guys have set up? Then also, maybe a suggestion for the committee to consider at a later time is would your group be willing to -- since you have established a reporting system, you obviously have taken a shot at trying to standardize or in some way uniformize, if I can use that word, the types of conditions that are being reported, and it may be that this may be a mechanism for the committee to utilize as we look towards how we're going to count these, are we going to be lumpers or splitters in these disorders. Then I have a comment after that.
But I think most people would be interested in, if they're not already aware of, your reporting system that just is up and running.

DR. THERRELL: I really would rather defer that to tomorrow, when I have a formal presentation first thing in the morning and discuss it in some detail then. I'd just say at this point we do have a formal system, and it is being participated in by almost every state. I'll elaborate more on that.

In terms of counting, lumping and splitting and that sort of thing, I think Dr. van Dyck is going to probably get into a discussion of what's been going on there lately. Of course, our group, being funded by HRSA, is always interested in the things HRSA is interested in and the things this committee is interested in.

DR. BECKER: That's fair. If I could make a comment, one of the questions I think Jennifer asked was some of the drivers to expansion. I can speak from at least one state's experience, that in 1998 we were doing five disorders at that time, which was competitive, comparable may be a better word, and we were considering adding CAH, CF, and biotinidase deficiency screening to our panel. Along about that time came some pressures to expand for disorders that we had to use MS/MS testing to accomplish. So our attention quickly turned towards accomplishing that task, which we did, phased it in like several other states have since done.

Then in 2003, we added screening for congenital adrenal hyperplasia. In 2004, we added screening for biotinidase deficiency. In 2005, we'll hopefully -- the fee and all the other things being approved -- add CF. So it's sort of deja vu. We're six years, seven years removed from what we wanted to do, but my point here is I think a lot of states have probably focused on MS/MS for probably what would be reasonably obvious reasons.

The other point is adding CF, biotinidase deficiency and congenital adrenal hyperplasia is on technology that's not MS/MS driven and usually requires -- although most labs probably have the capabilities, might have to acquire another couple of boxes. So there are some differences in what states are bringing on board, probably driven by the kinds of conditions that are being requested of them.

DR. THERRELL: Let me make one more comment. Dr. Howse, I said consumers, but I don't by any means mean to infer that consumers are the only ones. I have a slide that shows all the different contributing factors, and one time I gave that slide, and afterwards somebody came up from a professional group and said you have consumers but you don't have professionals. There are a lot of professional organizations that are also pressing to expand programs and change the direction of programs. So I don't want to overlook the professional groups.

DR. RINALDO: Of course, to me, perhaps a simpler answer to the question asked by Jennifer is that probably the main driver now is the maps that you see. It's becoming a standard of care. So once it was a few, then a third, more than half, and now we're approaching a point that perhaps three-quarters of the programs have expanded. Even if I agree, we probably can debate for years the issue of how we count and what we count. Still, with some variability on the margins, I think pretty much we know what the target is, and that's what I think is driving.

I think states do feel -- there are a number of states that clearly feel that they are behind, and their constituencies realize that probably their children or their babies are not offered the same service. Don't you agree that overall it's the big picture, one of the big drivers?

DR. THERRELL: Right. One of the things is it takes some money to start these programs up, usually. I mean, there are ways to do it where it doesn't take as much money, but it does take some start-up money. So the legislatures that control the purse strings don't often understand this, and they have to be educated too, and that takes professionals and consumers and a lot of people trying to help educate the legislatures. Once the legislature gets the picture, then they move forward usually.
DR. HOWELL: Dr. Telfair had a comment.

DR. TELFAIR: Thanks, Brad. I appreciate your presentation. One of the things that I think, if you could take a moment -- maybe you're going to plan to do this in one of your other presentations -- is a little bit about the relationship to follow up with this work. With a lot of screening that's going on, one of the things in terms of just getting the attention of those who have to make decisions is just the outcome of it in terms of the effectiveness. I wonder if you could speak to that relationship between what you presented and also what states may be doing to look at the follow-up issue.

DR. THERRELL: Well, the state programs have generally two aspects. One is an administrative/follow-up aspect, and the other is the laboratory. In the follow-up end of things, traditionally we haven't done very well long-term follow-up. We do short-term follow-up pretty well, and those are the numbers that get reported back to us, and they're the numbers that I'll talk to tomorrow about what's getting reported on our Internet system.

As you expand the program, you have to take care of follow-up because it's not good to do the testing and not be able to get the treatment and the diagnosis done. So follow-up is definitely an issue, and it generally costs in the neighborhood of the same thing it does to add the laboratory. When you increase the fee $10, usually $5 is laboratory and $5 is follow-up, if the fee even covers follow-up, because a lot of the fees in the states only cover laboratory, or if they cover follow-up it's done in some back-handed fashion where it may go into a general fund and then they have to compete for it.

So follow-up is a big issue, especially with funding, and it's an issue in terms of resources for these rare conditions. I think Dr. Puryear will discuss this as part of her look at the collaborative projects that HRSA is funding right now. So I don't want to get too much into that. I'll let her handle that when she gives her presentation.
DR. HOWELL: Does anyone want to comment more about the questions of how do we count? I mean, there's been a tremendous amount of discussion about that. Is it an issue that requires further discussion, or should we just have an understanding that many people count differently? Or is Peter going to tell us how we should count?
DR. VAN DYCK: Sorry, can't do that. Counting conditions has become a real issue, I think for everybody, and I guess I have been concerned that we're beginning to see the competitiveness among states and among testers for how many conditions are tested. So as we think about this more, it just seems like a good idea to try to arrive at some generally agreed upon way to count conditions that at least most of the community agrees to.

So with that in mind, we had a small group together about a month ago to at least begin this thought process and are preparing something that I hope we can produce for the committee for some comment

before the next meeting. It's just really something to get the thinking started, because eventually there is going to have to be a recommendation of some kind on how many conditions should be screened or what's recommended as a panel, and I think it's important to have some common definition for how we count.

DR. HOWELL: (Inaudible.)

DR. VAN DYCK: I would not call it a report. All I'd call it is a thought piece to put the issues in front of the committee to begin a discussion on how we might want to approach it.

DR. HOWELL: So we anticipate that it will be available for the next meeting, perhaps?

DR. VAN DYCK: I would think so.

DR. HOWELL: Good.

I would seem to me that it would be helpful to have a general understanding of that, because I think you're correct, that the competitive juices flow when you're trying to count conditions, and we agree that many people do the same test and have very different numbers that they count.
Bill, you had a comment.

DR. BECKER: Yes, just one more point I think that at least I gleaned from Brad's presentation is something that I think we need to -- I guess I'll remember and it will probably come as a by-product of what Peter's work will be presented later. Right now, there is no requirement for the states to report conditions to any group, whether it's Brad's database or otherwise, and I think that's something we're going to have to probably deal with.

DR. BOYLE: Brad, I was going to ask you maybe a little bit of an unfair question, but this goes back to what Jennifer brought up earlier. Obviously, one of the charges for the committee over the next two days is to think about subcommittees, and you've worked in this area for a long time and have, I'm sure, a lot of thoughts about that. Maybe you can provide some -- and you don't have to do it now, you could do it at your next presentation, but provide some guidance to the committee in terms of what your thoughts of what the most appropriate subcommittee structure might be for this, what are the real pressing issues and how can this committee be most helpful in that regard.

DR. THERRELL: You're right, I'll wait. Actually, in my next presentation I give you some challenges at the end, and I think this will fit in well there.
With respect to counting conditions, I will say that March of Dimes has been a plus and a minus in this respect. March of Dimes has traditionally had a report card for the states, and this became nationally obvious when Dr. Howse was on TV a few days during the summer, and states were very interested in being scored high by March of Dimes. At that point the March of Dimes was scoring states whether they did 10 disorders or not.

After your last meeting, March of Dimes quickly went to a scoring or announced that they would be scoring for more disorders, including the core and the secondary. So states have begun looking at this numbering thing. After the presentation Dr. Howse gave on TV, for instance, I got calls from the people in Minnesota who were very upset because the data had come from us to March of Dimes and Minnesota wasn't counted as doing the 10, but Minnesota was doing 30-plus. Well, the reason they were counted in the 10 was because they weren't doing biotinidase, and as you saw they now are doing biotinidase so they would make the 10. At that point they would make the 30 but they wouldn't make the 10. So it's that kind of issue that's been going on in terms of competitiveness and in terms of scoring.

But it's been very nice, actually, to have an outside group scoring states in some respects, because the states are looking at that and they're saying, well, maybe if everybody else is doing this, we should be doing it, and it gets the juices flowing within the states and helps kind of drive some of those issues.

DR. HOWELL: I think Jennifer has a comment on that.

DR. HOWSE: So, guilty as charged.

(Laughter.)

DR. HOWSE: I think that we intend to continue with the report card approach and really use that as a way to continue to focus national attention on the disparities between states on which newborns get which tests.

But circling around and connecting directly with what Dr. van Dyck brought up is the subject of what is the nomenclature that's going to be used for the tests, and that could be a very long discussion. That could be a discussion where consensus is perhaps not achieved for the reasons that you so clearly brought out. So just to put kind of a line in the sand on that one, I think what our organization believes at this point is that the report from the American College of Medical Genetics is as good a way as any, and also represents reasonable professional consensus about how to name the tests and call the tests and list the tests. Again, we feel it's very important to get that recommendation or that report into the public domain so that that can be the starting place for activity that is already so far down the road on a state by state basis, and to get that report into the public domain, to put the list out that's been the subject of years of work by very respected medical professionals -- no list will be perfect. There's always going to be arguments for the reasons that were well laid out, but we feel that that's a very, very important starting point and plenty good enough to move the issue ahead.
So, guilty as charged.

DR. HOWELL: Peter, you had some comments.

DR. VAN DYCK: Well, just to back up a second, there have been a couple of comments about the subcommittees, and that is one of the purposes of this meeting, I think. I just want to say that this afternoon I'm going to present in my presentation right during the start of our business meeting a list of the issues put together by, again, a small group on our staff that we feel should be considered by the committee. So it's just going to be a short presentation trying to get all the items on the table for you all, and then you can arrange and subcommittee them anyway you choose, and I think that would be a good point for Brad and others to suggest. But we'll try to lay out a scheme that at least brings to focus all the issues that we have thought of related to newborn screening.

DR. HOWELL: Brad?

DR. THERRELL: Just one further aside. You saw the press release I had from Mr. Pataki in New York. One of the things he did was he said New York now screens for 44, the number-one free program in the country, and the press immediately jumped on that. What they were missing was the word "free" because New York doesn't charge a fee, so they're one of five states not charging a fee and only one of those five that can say that. So that's another issue that gets brought up.

DR. RINALDO: Brad, I'm glad you brought that up, because in my mind this issue of counting has been really blown a little bit out of proportion, and I really see it in your presentation. You said this seems almost as a consequence of marketing strategies. But in reality it really seems to me representative of state newborn screening programs when placed with a microphone in front of their mouth. They can't resist to try to pretend to be number one. It must be the sport season or the playoff influencing things. But the tragedy, if any -- well, not tragedy. The serious consequence of continuing thinking along these lines it seems to be to hijack the concept of quality. But the quality of a program now rests with the number of conditions, so the more, the merrier.
That is really a major sort of step in a negative direction. Do you agree with that?

DR. THERRELL: Yes, I do agree with that, and that's why states have been slow to increase their numbers, I think, because they're very much interested in quality. Now that they're jumping into things,

sometimes the quality does suffer. I agree that states market just like private companies market, so I say marketing in the generic sense.

DR. HOWELL: Are there further comments on the subject?
(No response.)

DR. HOWELL: I think that the competition between the states and the report card that Jennifer talked about has been very effective, however, in moving the process along, and I think particularly since we're meeting in the Ronald Reagan Building, we would have to say something good about the free market and competition.

(Laughter.)

DR. HOWELL: On that note, why don't we take a break? We're actually right on schedule, and we'll resume in just a bit with Dr. Groft's presentation.

(Recess.)

DR. HOWELL: Everybody is here at the table. Thank you very much.
Dr. van Dyck had to go to a reception with the Secretary, Secretary Thompson, who is apparently meeting with the leaders of HRSA today as a departure effort, but he will be back very soon.

It's my pleasure to introduce to you Dr. Steve Groft, who is Director of the Office of Rare Diseases at the NIH. Again, one of the key things that we really want to focus on is following up these persons who screen positive, not only defining the best treatments and so forth, but also they need to be connected into a research environment to find out new treatments and look at long-term follow-up and so forth. Dr. Groft's group in rare diseases has been very active in this area.
Steve?

DR. GROFT: Thank you very much, Dr. Howell.

I hate to interrupt a good time, so I hope everyone enjoyed their conversations and can continue them at lunchtime.
Thank you very much for the opportunity to come to the meeting and discuss some aspects of our program, what we're currently doing, as well as some other activities I think with direct implications with respect to genetic testing. It's sort of an exciting time for many of us who waited for a number of years to start working to implement many of the ideas that have been discussed for many, many years.

I'll see if we can get all the slides working together here.

In 2002, the Office was given a little bit different mandate. In fact, we had been working under administrative activities at the NIH, and in 2002, with the assistance of the patient advocacy groups, we were able to get legislation passed that mandated certain activities within our office, and there was an emphasis on clinical research. You can see the various major sections that we had to devote our resources to. With this mandate came an increase in our budget, and we currently are funded at the level of $15.5 million. That's an "M", as opposed to some of the other "B"s that some of the institutes have.

But I think one of the very nice things about our office, where we're located within the Office of the Director at NIH, is that we can serve as a stimulus to initiatives that need to be addressed. It seems like, with respect to the rare diseases, we have so many willing partners both within the NIH structure and from the other agencies and departments in the government. So there never is a lack of partnership on different activities, and that's what we try to focus on, initiating new activities, providing seed money for other activities that will get things started, and then try to reach into the deeper pockets for additional funds. I think you'll see that throughout our entire program that's been our method of operation. It's about the best that we are able to do.

If I can just for a moment define a rare disease, it was back in the '80s when the original Orphan Drug Act was passed, and with amendments to that it defined a rare disease as one with a prevalence less than 200,000 here in the United States. So when you think about it, if you have a disease with that many people, it's not uncommon but not as many as a more common disorder, but certainly enough that a company probably can make a profit, and that was one of the major concerns initially, that a company would not develop a product without making a profit. So I think they put the number at about one-tenth of 1 percent of the United States population at that point in time.

The Act did, of course, give you certain responsibilities, and it usually requires writing reports. So we've just completed our first biennial report, and it will soon go up on the web as soon as clearance is obtained through the NIH Director's Office, with the assistance of the institutes. We do give the institutes quite the opportunity to expand on their programs and present it to the public with a focus on the rare disorders, and all this information is readily available on the website.

We were given the responsibility of preparing a couple of reports. One is an annual report on the advances, so we tried to fold that into the biennial report this year. It also gave us the mandate to develop an information center with respect to the rare diseases, and we expanded that to include genetic and rare disorders, and we established that in 2001. We just received our 10,000th inquiry in December of this year for 3,000 different rare disorders. So there's quite a bit of need for information, as you all know.

I guess the issue is when we receive the questions, many times the people have not been able to obtain the information elsewhere. So a lot of times it's a customized search. We're working on the process of having computerized responses so we can just update as we go along from inquiry to inquiry. Last year we also expanded the services to include Spanish translation and available to receive inquiries in Spanish from the public.

The Office has been focusing on a number of activities. Before 2002 we were looking at developing information and dissemination with respect to the rare and genetic disorders, and when we started receiving the increase in our budget we looked at different activities and we were able to implement an intramural research program, expand the extramural research program, and I'll talk about this.

If you didn't get slides, there are copies back there for those in the audience who may not have received a copy of these slides, if you need them.
So we've initiated these programs. We've established a trans-NIH Working Group on Rare Diseases. So for the first time we've been able to get all the institute people together to focus on rare diseases research. So it's a real nice working group, and we've had two meetings, and I'll explain a little bit more about some of the activities there.

Just first our extramural program. We've done a number of scientific conferences. As I mentioned, we have been in operation since about 1993 administratively, and since 1995 we've had 500 scientific conferences on different rare diseases. All of these conferences are done in conjunction with at least one of the research institutes at NIH. So there's a great deal of collaboration. We look for international researchers, national researchers, try to get the Food and Drug Administration and any other group who has an interest, including industry, patient advocacy groups. So it's been a real nice program for the Office.

This year we've expanded to do three review cycles. We had our first review cycle, and we provide support for 60 scientific conferences this year, and we anticipate doing about 100 of these in the current year. So it's quite a bit of work for the Office to track these and really to be involved in the development of the agenda and the planning for these meetings.

We established the Clinical Research Network, and I'll talk about that in a little bit.

We've been working with the patient advocacy groups. Over the years we noticed a need for the patient advocacy groups to have a better understanding of how the NIH works, how the Food and Drug Administration's Office of Orphan Products works. So we have these weekend seminars where we bring together the leadership of patient advocacy groups. We have a third one this year in Philadelphia. We limit it to about 50 people per meeting, and we have a series of presentations with question and answer periods to try to get them to become better informed, individuals who actually are the interface with the public for their rare disease, and with health professionals. That has worked out very well, and we're looking forward to continuing that process as well.

We've developed a number of research initiatives, again very small, but we're able to contribute some money to these demonstration or pilot projects. We've had a lot of interest in lysosomal storage diseases, and you can see the clinical trials planning grant, all major initiatives to focus on rare diseases research, generally areas that need to be completed if we're going to have research moving forward to the stage where we're talking about the development of a product. We try to focus on those, and we even have a training program in genomics and proteomics to study rare disorders with the Human Genome Institute.

In the intramural research program, Bill Gahl is the director. He's doing this in conjunction with his responsibility with the National Human Genome Research Institute. Again, this is almost a test bed for projects that we like to see eventually be expanded to the extramural program, and then other areas, depending on the staff, that we're able to become involved with us. You can see some of the programs. Bill has the training program for biochemical geneticists, a major need with respect to the future. We've initiated a so-called pilot project, if you will, with patient travel, helping individuals get to the research and treatment sites, and we're looking to expand this in the future to provide support for an infrastructure. It's usually free travel, and it can be through commercial flights, corporate jets, and through private pilots to get to and from research and treatment sites with Mercy Medical Airlift. Then we also work with the Hospitality House Association to provide lodging.

Bill has developed a protocol for undiagnosed diseases to bring patients in after reviewing the records, to get a focus and bring a group of researchers together to sort of brainstorm what might be wrong with a patient who has not been able to obtain a diagnosis. This has been a slowly developing program, but we look forward to continued expansion of this.

Bill has also worked on the development of genetic tests for four disorders this year, and we hope to expand to 10, trying to identify the needs of the research community. With our activity with respect to genetic testing that we'll focus on in a little bit, again it's a test area to find out where the problems are. So if we go into an extramural research program and a translation from research to the clinical services or clinical laboratories to meet the requirements of CLIA certification, CLIA laboratories, we hope to use the experience from this, and we've gained a great deal of acceptance of this possibility with the other institutes.

Our last intramural program is our bench to bedside awards that we make throughout the NIH structure, and this requires a clinical and basic research component, as well as at least two institutes involved in a research project.

Just to briefly highlight why we provide a lot of focus on the scientific conferences, you can see some of the outcomes where we talk about establishing research priorities, developing program announcements. Many of these initiatives are what is needed to get research moving on so many of the rare disorders. I think with so many patients located at a distance, and even very few investigators, we need to have a focus on bringing the people together willing to identify the research agenda and help move research forward. This has been a very nice program and we think very successful to utilize as a mechanism to stimulate research with the rare diseases.
Even with this, we work with the patient advocacy groups in the development of the agenda and definitely have their presence, along with the international investigators, investigators from the United States as well.

Here is one of our most recent activities, the development of the rare diseases clinical research network. Again, it's a model. With patients scattered throughout the country, throughout the world, we observed that there was a need for a systematic collection of clinical information. It's difficult, as many of you know, to gain enough patients together at one place for research to actually move forward with respect to the rare diseases. There is a copy of all of the consortia I think within your background book under Tab 6. All of the information, too, if any of you would like to refer to it later on, is prominently listed within our website. I'll give you that information as well, too.
But again, looking at biomarkers, just many things that need to be done with respect to rare diseases. So this was a nice model to see what we might be able to accomplish in an organized and systematic fashion, and then translate that over to many of the other rare diseases. What we're noticing as we go along, we're getting many requests from patient advocacy groups hoping to join us, but it's just not possible to do. But I think what comes out of the network will be information to guide many, many research activities, and there will be a model for so many of the rare disorders.

When we were looking at this, one of the major deficiencies in rare diseases is this lack of longitudinal natural history of the disorders. We don't know exactly how the disease progresses over a number of patients. So we required a longitudinal network history component to the research application that came in in the proposal. So it is a little bit different approach, and it was one of the areas where the research investigators were telling us all along that they were unable to obtain funding, so we thought we'd try something a little bit different here and require this as part of it.

We also required a commitment and participation of the patient advocacy groups for whatever diseases are included under the grouping of rare diseases, and they did have to include at least two different diseases. Many of the actual awards that were made included several disorders, and from this whole network of 10 consortia there are 70 sites located throughout the United States, affecting about 50 rare disorders and 30 patient advocacy groups. So we are trying to interlink, and we hope that what comes out of this is a referral of patients from site to site, even if it's not within their primary working area, but to facilitate travel and to facilitate the access to the clinical trials that will be started.

To help with all of this is our data and technology coordinating center. Jeff Fisher in Tampa is leading this, and it's quite a bit of work to get this design of clinical protocols and to coordinate data collection. I think anyone who has done a multi-center study, this is what we're looking at for a number of diseases. So it's quite a bit of work that he has to do to get the investigators to work together. We've got a web-based recruitment tool that is available to the public, and so we are looking at this as a model for more rare diseases, and again I think looking at where we will go in three or four years. We would really like to have this website be extended to many, many more rare disorders, even those outside of the network that we are putting together.

I think it will be of some interest to see how it all develops and if we're able to really do this. In order to effect this network, as was mentioned earlier, you do need subcommittees, and these are the various working group subcommittee types that we've had to put together looking at standards and terminology, how do we track specimen tissue, how do we even track data from site to site and into the data coordinating center and back out. We've established a Data Safety and Monitoring Board that will meet for the first time at the end of the month, and we have 12 protocols that will come forward, five interventions, and I think there are six other studies that will be involving longitudinal and natural history type studies.

Here is the list of the various disorders. Again, it's quite extensive, and it's highlighted prominently in your book as well as on the website, so I won't spend too much time with this.

We did establish a coalition of patient advocacy groups. I believe the feeling has always been that when research projects are initiated, the patient advocacy groups and voluntary patient organizations just are not listened to. They have so much to offer that we made them an actual voting member on the steering committee, and we have our first meeting of the group. You can see some of the activities that they have an interest in, rare diseases emergency room treatment, critical care treatment guidelines, looking at best

practices treatment guidelines. These are some very interesting concepts that for the rare disorders just have not been done. So we would like to initiate this, and we're going to try a couple of these activities to see what we're able to accomplish on different rare diseases, again a model for others as we move forward.

Here is the trans-NIH working group, some of the activities that we've been really able to work together on. Again, for the first time, we've been able to bring all of the institutes to focus on the rare diseases. Again, we've had two meetings, and some of the activities, in order to foster the collaboration and coordination of research and other activities within the NIH, we thought this would be the best mechanism in several of the areas -- development of diagnostic genetic tests, and collection, storage and distribution of biomaterials for research.

What we found is that there are many activities going on, but it really isn't a coordinated effort. So we've established a couple of working groups to discuss these issues, and then to report back and start some type of implementation that we can begin to bring some order to many of the rare disease resources for the research community, and as well for patients and clinicians.

Recently you may have heard some things about promoting quality genetic testing. Again, this is a website that has the presentations and a report on genetic testing. We sponsored a conference last spring with CDC, HRSA, CMS, a lot of the other acronyms, the alphabet groups here within the government, as well as the American Society of Human Genetics, American College of Medical Genetics, the Genetic Alliance and several of the other groups that are involved in all aspects of genetic testing, trying to identify issues and needs that we felt needed to be addressed, many of us felt needed to be addressed.

So these issues were brought out, and we will be discussing this at a rather large meeting in the fall of this year to roll out, more or less, some of the plans and possibilities from the government, from the private sector. Again, it's a need that I think has been identified to guarantee that there is, first, a translation from the research laboratories to the clinical services, and then adequate access to these genetic tests, with some assurance that they are going to be correct.

So we'll be continuing to work on this. Some of the issues that need to be discussed -- and we will continue to discuss and come up with some plans -- are international testing regulations and quality of global testing services for genetic disorders, appropriate result interpretation with adequate patient counseling, and then again we're trying to foster the partnerships and networks to improve research translation and data sharing with the research and clinical laboratories, and then among the research investigators and clinical laboratories, patient groups, clinicians and payers.

As has been mentioned, things just don't move without this collaboration and coordination. So we're hoping that we can initiate some good activities and really have it work together.

Again, I talked about facilitating research, and some of the other areas that will be discussed are cost of test development, reimbursement, liability concerns for the test, and then quality assurance, especially as the tests are administered in prenatal diagnosis, and that we get some service back-up when necessary and have comparisons between laboratories to provide some measure of assurance that they are correct.

And just some information, contact information for the Office and the Rare Disease Information Center, as well as information on our website that we try to provide, so I won't spend any time on these two. Then finally, the contact information for the Office.

Thank you very much.

Michele, are you presenting, or should I take questions?

DR. LLOYD-PURYEAR: We're going to let you answer questions.

DR. GROFT: Okay.

DR. LLOYD-PURYEAR: The Chair has stepped out. I'm not sure what the protocol is.
Does anybody have any questions for Dr. Groft?
And thank you, Steve.

DR. HOWSE: Dr. Groft, could you just outline again the budget for your office and sort of give us a sense for -- this is a very well thought through comprehensive program. Could you just give us a sense of the financing that's available for some of the major aspects of your office?

DR. GROFT: The budget is currently $15.5 million, and I don't anticipate a great increase. But what we've tried to do is, for example with the scientific conferences, we provide up to $25,000 per conference, and hopefully that is matched at least by the institutes, and generally it is. For the network, the coalition, the research coalition, it's approximately $1.25 million total cost per coalition, and again we have 10 of those and one data and technology coordinating center that's over $2 million.
Again, we originally thought we could -- we hadn't sufficient funds to do four, but with the cooperation of several of the institutes that I identified there, we increased to seven, and then we had a small increase in our budget last year that enabled us to do 10, to increase to 10. We had a tremendous amount of interest when we published the RFA for the number of academic centers that brought forth applications, many more than what we ever could fund, unfortunately. In fact, we probably are at maybe 15 percent of the applications that we're able to fund.

DR. HOWSE: Fifteen?

DR. GROFT: Fifteen percent. Not a lot. At that point, the (inaudible) is a little bit more flush with their budget. So even on the activities with respect to the genetic testing, traditionally what we try to do is identify issues and then try to refocus and reorganize our budget to provide resources that are needed to start an activity or to gain partnership with the other institutes and centers or other government groups to do this. It's a lot of brokering. We just try to work through the issues.
Is that okay?

DR. HOWSE: Yes.

DR. HOWELL: Dr. Dougherty?

DR. DOUGHERTY: So a follow-up question to Jennifer's. Do you expect to be able to issue another funding announcement any time in the future, or is all the money in your budget going toward continuing costs?

DR. GROFT: Yes, yes to your last question, and I don't think so for the first question, unfortunately. I don't anticipate a great increase in the budget for our office, but we're hoping that as we develop this, what we're noticing is that some of the organizations and academic centers that submitted applications or proposals and were not funded, they're continuing and they're strengthening their relationships with each other, and we're now looking to the institutes to find other funding mechanisms that might be able to be employed, some program project grants and others that we can develop.

You saw the emphasis on lysosomal storage disorders. We were able to develop an RFA with that to get out. So I think as we hear things and as we hold more conferences, the follow-up for so many of the conferences is this stimulation of research on the rare disorders. But it would be nice if we could do about 100 of these centers. I think that would be super.

But again, the institutes themselves are funding a lot of research with rare diseases. I think there's a misperception that the NIH and the institutes don't fund a lot of research with rare disorders, when indeed we really do when you look through the portfolios. It's just that when you're looking at 6,000 or 7,000 rare disorders, 5,000 or whatever -- we really don't have the accurate numbers -- it's a wide distribution of many, many diseases throughout many institutes. But when you put the whole portfolio together -- I've done searches on our one network to track grants, and when you put in the terms and get the printouts for one page, it comes out a couple of feet, two or three feet. It's a lot, having read through all of them. I don't want to be too compulsive, but it's actually good reading.

DR. BOYLE: I was just going to mention that I think that the Clinical Research Network really provides for opportunity to do a lot of what I think we've discussed in this committee in terms of looking at the natural history, the impact of clinical treatment on many of the rare disorders that are in the purview for newborn screening. Even some seed funding to bring together -- and I'm just speaking to the committee here -- to bring together all of the research that NIH and perhaps other agencies fund and allows that sort of unifying framework for it is really terrific.

So I applaud you, and I also think that given your budget is only $15.5 million, we don't need a lot of infrastructure here.

DR. GROFT: No. A lot of the infrastructure already exists. It's a matter of employing it appropriately and then extending the areas that need to be looked at. In fact, we've had a number of meetings with Michele and her group just to talk about how we can work together, especially with her network and our network. So we're looking at ways how we can integrate both groups even more, and I think we can be effective.

But again, we couldn't do any of this without the institutes. It requires so much money. Dr. Alexander is here, and they've been extraordinarily supportive, as have others. So it's been super.

DR. HOWELL: Any further comments?

Bill has some comments.

DR. BECKER: Thanks. That was a great presentation, and I realize the focus of your program is mostly to develop clinical investigators, but I'm going to assume that you probably don't -- or maybe I'll just ask. Do you have any of your RFAs that interact with, link with, utilize the resources from traditional newborn screening programs right now?

DR. GROFT: Not right now, no.

DR. BECKER: That was my suspicion. It may be that establishing a link -- obviously, as programs are more and more successful, as we've heard already this morning, and screening for more and more of these rare disorders, that there's going to be a repository of samples -- DNA, serum -- that could be utilized for your clinical investigators, and vice versa. The spinoff of the development of some of your RFAs could spin back into clinical use or what I consider use for the newborn screening laboratories as well.

DR. GROFT: I think that's one of the things we'd like to do, that issue of biospecimen collection, storage and distribution. We really would like to get a better hold on that and make these services that are already in existence more readily available and known to the research community, as well as to the patient community who want to donate tissue. I mean, they are such willing partners to contribute anything while they're alive and after they die that I think we just have to make it easier for them to gain access.

I can't tell you the number of times we get calls from patients and their families who are dying and who want to contribute but they don't know how to get to it. This has been a persistent problem for many years. So I think we're going to try to work with a number of the institutes to really develop this a little bit

better and make it readily available.

DR. HOWELL: I think that your group has done a remarkable job, and I think having a center of expertise on these rare conditions when they are identified through newborn screening, that they can be communicating about treatments and so forth is invaluable. I think your goal of having 100 centers is modest.

DR. GROFT: Reality comes into play every now and then.

DR. HOWELL: Well, hopefully not too often.

Thank you very much, Steve, for that excellent presentation.

We've heard now about the research efforts, and Dr. Puryear is now going to tell us about HRSA's newly funded Regional Genetics and Newborn Screening Collaboratives, which again I think will have a potentially very excellent role in the infrastructure program.
Michele?

DR. LLOYD-PURYEAR: This is the service part of the infrastructure. When you look at our budget, there's a considerable contrast in what's provided for research. We're working with a budget of $4 million, as opposed to $15.5, for the Regional Collaboratives.

Anyway, the Regional Collaboratives are one of our newest initiatives, begun in 2004. We created this initiative to respond in part to the heritable disorders program legislation, which is also legislation, as a reminder, that created this advisory committee.

I first would like to place the initiative that was developed in the context or the framework of the mission of the Maternal and Child Health Bureau, and also our legislation. The mission of the Bureau is to provide national leadership and to work in partnership with states, communities, public/private partners and families to strengthen maternal and child health infrastructure, show the availability and use of medical homes, and build the knowledge and human resources in order to assure continued improvement in the health and safety and well-being of the maternal and child health population.

That population has been interpreted to include newborns, children, adolescents, women and mothers, and women of non-childbearing years also, and fathers.
Our legislation indicates that we are to fund activities to provide for what's called Special Projects of Regional or National Significance, or SPRANS; for research and training for maternal and child health and children with special health care needs; for genetic disease testing, counseling and information development and dissemination programs; for grants, including funding for comprehensive hemophilia diagnostic treatment centers relating to hemophilia without regard to age; and for the screening of newborns for sickle cell anemia and other genetic disorders and follow-up services.

This was actually, until the heritable disorders program, the only federal legislation for funding for genetic services.

We've organized our description of maternal and child health services into a pyramid, about which Peter will go into more detail later on this afternoon. We provide funding for all layers of the pyramid, but our primary focus has been on infrastructure building services with the idea that this is the essential foundation for the other levels of the pyramid, and this includes such things as need assessments, evaluation planning, policy development such as the Newborn Screening Task Force Report that we did with the Academy of Pediatrics and other federal agencies, standards development, information systems development, and you'll hear more about that later on.
We also, when we created the Regional Collaboratives Initiative, we examined the current landscape, and we've heard over the last two committee meetings about the changing technologies and many of the challenges it brings, moving for example from testing for single disorders to multiplex technology, where you're testing for more than one disorder at a time, and some of the challenges that brings.

Most of the disorders that are tested for are very rare disorders, and Steve has talked about it both in this country and internationally. There are generally very few providers with the required expertise. At the first meeting the committee heard from Dr. Howell about the new technology on the horizon bringing additional challenges.
The legislation for the heritable disorders program was enacted in 2000, and in 2003 the committee was recreated at the request of Congress. They asked us in our appropriations language to begin implementation of this program, and we began with the creation for this committee. Then more recently we had to consider the recommendations from the American College of Medical Genetics Newborn Screening Expert Group.

Last fiscal year our appropriations language, Congress indicated that we designate funds, and they indicated that we designate $2 million to implement the heritable disorders program. We matched that with an additional $2 million. I want to go through the legislation as a reminder about what it is, but also as a reminder that this committee, one of its functions within the legislation but also in the charter is to advise on this grant program.

So the legislation indicates that the Secretary shall award grants to eligible entities to enhance, improve or expand the ability of state and local public health agencies to provide screening, counseling or health care services to newborns and children having or at risk for heritable disorders. The legislation indicates that funds be used to establish, expand or improve systems or programs to provide screening, counseling, testing or specialty services; to establish, expand or improve programs or services to reduce mortality and morbidity; and to establish, expand or improve systems or programs to provide the information and counseling on available therapies.
In addition, to improve the access of medically underserved populations to these services; and to conduct such other activities as may be necessary to enable newborns and children having or at risk for heritable disorders to receive those services, and this is regardless of income, race, color, religion, sex, national origin, age, or disability. So it's pretty all-encompassing, but it tends to focus on the service infrastructure for newborn screening programs, although the last indication of what funds should be used for is not so specific and is more general and allows other aspects of the program to be addressed.

So we began last year with developing guidance, a request for applications for seven Regional Collaborative Groups and one National Coordinating Center. The country was divided up into seven regional groupings based on birth rate, to equalize birth rate, geographical proximity, and a recognition of some preexisting working relationships the regions had already established.

The guidance requested activities or a proposal for activities specifically to enhance and support the genetics and newborn screening capacity of states, and those activities to address maldistribution of genetic resources -- that was of primary interest in the initiative -- to promote translation of genetic medicine into public health and health care services; and ultimately to shift services into local communities.

We asked for specific description proposals of activities that would enhance screening and follow-up services, augment capacity needs, specifically training and education needs of the region, to describe activities to strengthen linkages to the medical home and tertiary care providers together, with the idea -- and Coleen spoke about this in the last question and answer period -- for developing paradigms of relationships, new paradigms to be able to follow a child identified to look at the long-term health outcomes of children. So this will require a linkage between medical homes, tertiary care and the newborn screening programs that we don't think exists right now.
We wanted activities to strengthen genetic counseling services and activities to enhance communication both between the regions and within the region, and education to family and health care practitioners and other general forms of information sharing.

We asked that the applicant have the willingness to serve as a regional center for that region, and the region could be virtual. It didn't need to be located in one place.
We wanted the activities that were described to be based on the capacity needed in the region, and

several of the regions, the states within the regions had actually participated in a previous initiative of ours that we began about five years ago of funding the development of state genetics plans. Many of the proposals where that happened were based on those needs assessments. So that was actually a very valuable stepping-up of a previous initiative.

We asked that the regional center organizationally represent a variety of collaborative partnerships, especially between the public health agencies and clinical community providers.

We also asked, if they existed in the region, for the region to have a willingness to have partnerships with the CDC's Centers of Excellence for Birth Defects Prevention Research and NIH's Rare Diseases Clinical Research Network.

We also pointed to some required endpoints within the first three years of these cooperative agreements. Because some had already participated in genetics planning activities previous to the launch of this initiative, some were farther ahead, but a few, probably half of the regions had not been a part of any of the previous development of state genetics plans. So we began with the development of a regional coordinating plan to address specifically the maldistribution of genetic and newborn screening services and expertise.

We wanted them to also present some practice models and materials that would be needed for optimal diagnosis, follow-up and management of the children identified within the region, and by year 2 to develop a strategy to implement those practice models.

In year 3 we want the collaborative to demonstrate public and private regional partnerships and relationships that represent the variety of health systems organizations within all states within the region. We required at least four states within the region to be part of this first part of the initiative. At the end of the third year, we want all states to be represented. We know we can't require that and we can't force a state to participate, but we want that outreach to be given.

We also wanted to see the inclusion of community health centers, health care insurers in these partnerships, health maintenance organizations, the state-based primary care organizations, and academic institutions. Of course, we emphasize the importance of partnerships with the state genetics and newborn screening advisory committees within the defined regions.

The regional abstracts are in your briefing book. These are the states that are in the different regions. They're covered on the next two slides, and I'm not going to present those in detail.

I'm going to go through on the next seven slides some of the activities that the regions have proposed. The first is Region 1, which is the New England Genetics and Newborn Screening Collaborative. This is based on a preexisting relationship with the New England Regional Genetics Group, or NERGG. The project director is Tom Brewster. The co-project director is Ann Cuomo and Susan Washburn. They proposed three primary activities, and that was to enhance collaboration through communication -- they're using a website methodology for that. They also want to enhance and improve current practice models. They're developing some best practice models using what they have conceptualized as an equality of access to newborn screening model. They're developing a geographic and epidemiologic approach to assessing their capacity needs of the region. They have a proposal to improve educational opportunities within the region. This includes the development of a uniform management guidance for treatment, and an educational committee is developing programs to improve the genetic literacy of the population in that region.

Region 2 sits at the New York State Department of Health. The director is Ken Pass. Their primary activities will be the development of a regional coordinating plan. This will be providing local solutions to barriers to access to specialty care for children with heritable disorders. One of their special projects if the development of a teratogen hotline. They also will be funding a series of state-specific projects which they're terming special regional projects. Many of the Regional Collaboratives have a special fund for state or small projects.

To address particular problems in newborn screening, they're working with Massachusetts to develop an emergency back-up system for newborn screening, and they're proposing to standardize newborn screening throughout the region. They also have an educational proposal that's directed towards providers, payers, patients and families.

Region 3, the Southeastern Regional Genetics Group, or SRGG, has two project directors. David Ledbetter is in Georgia and -- I know I'm going to say his name wrong -- Jess Thoene is in Louisiana. They're spending a significant first year identifying gaps in genetic services and conducting a needs assessment. Also, they have a significant project, a telecommunications project that connects states' academic and public health representatives to each other to create an ongoing methodology of regional communication.

Region 4 sits in Michigan Public Health Institute. The project director is Cynthia Cameron. They have a three-cluster area of work proposed. One is around newborn screening specifically with tandem mass spectrometry, again like other regions to achieve uniformity of testing at the testing panel within the region, and also to improve the analytical performance within the region in a uniform manner.

They have a specific project to look at inequities in access to services, and they're proposing some different methodologies to achieve that, to address the geographical maldistribution and looking at telemedicine, long-distance consultation, and the use of satellite clinics. They also have a public health infrastructure which is developing a practice model for optimal diagnosis, follow-up and management for the children that are identified with heritable disorders and birth defects.

Region 5 sits at Oklahoma Health Sciences Center. The project director is John Mulvahill. Their first year is focused on developing regional infrastructure to address communication, education and resource needs of the region. They also over the first year will be developing what they call the Heartland Regional Genetics Strategic Plan. They propose several special smaller regional projects to fund, and a primary focus of their first year will be identifying gaps in service and education.

The Mountain States Genetics Foundation is Region 6. The project director is Joyce Hooker. She's actually here today. They moved to establish the Mountain States Genetics Regional Center. A great deal of their activities in the first year are conducting a regional needs assessment, and upon that developing a regional plan for collaborative genetics activities.

Finally, the last region, Region 7, sits in the State Department of Health in Hawaii. The project directors are Sylvia Au, who is from Hawaii, and Kerry Sylvey, who is in Oregon. They already had the beginning of a regional plan. Many of the states in that region had already been part of a state genetics planning process, so they wanted to move to actually implement a practice model that they had already begun to develop across the region to focus on improving access to specialty metabolic genetic services and primary care.

For the states in the region that had not been a part of the initial planning process, they want to work with those states to conduct needs assessment to identify activities to increase the capacity of the other states, such as Alaska and Idaho, to increase the capacity for genetic services in those states.

We also funded a National Coordinating Center, as I indicated. The American College of Medical Genetics was a successful applicant. The director is Michael Watson. The primary focus of the National Coordinating Center is to support the regional collaborative efforts to identify issues specific to the utilization of genetic and newborn screening services at all levels.

Some of the activities will focus on minimizing duplication of efforts, identifying best practices developed by the regions -- many of those best practice models will be examined -- to further information exchange and professional collaboration between the regions, but also between national organizations that are a part of the ACMG effort or the National Coordinating Center effort, and to maximize interregional collaboration. One example that's given is to have language and terminology compatibility across the country.

Some of the plans that the coordinating center proposed would be to develop networks of centers of genetic services, or COGs, with primary care providers; to facilitate data collection, collaborating with the NIH rare disease centers and the CDC genomics centers; and to involve national programs to which ACMG already has relationships, such as the Academy of Pediatrics and the Academy of Family Physicians, to address CPT code development, but also work with organizations such as JCAHO to bring some uniformity of practice within hospitals for newborn screening programs, for the process of newborn screening sample-taking.

They also propose some information-sharing projects especially with those with overlapping interests around such things as telemedicine, for example.
We examined the applications of the Regional Collaboratives and with the National Coordinating Center identified several areas of shared need. Communication methodologies are a big shared need, ranging from videoconferencing to telemedicine, setting up practice models and practice relationships with genetics networks and the use of interstate satellites have been some of the proposals throughout the applications. Information management in general, but also how do you manage web-based clinical management systems suitable for telemedicine, or even for the satellite clinics.

Reimbursement is an issue. It's always an issue, but it's a specific issue when you're trying to work across state lines or a long distance, for example, for reimbursement for telephone consultation.

Evaluation methodologies were areas of shared need. Expansion of newborn screening, and therefore also the needed expansion of follow-up service infrastructure was an identified need. Specific regulation and legislation needed to be addressed to allow interstate licensing to address liability concerns across state borders. Financing has always been a need and is a specific need when you're calling for the expansion of newborn screening, but also expansion of the service infrastructure, and an expansion of relationships, developing new paradigms of working relationships.

They all pointed to a need in how to expand access to genetic services, specifically pointing to the need for training of geneticists, but also the training of primary care providers in the field of genetic medicine. There was a need to increase the diversity of trainees, and also a mechanism to more systematically address the geographical maldistribution.

The coordinating center has proposed these partners, the National Conference of State Legislators, American Academy of Pediatrics and the Association of State and Territorial Health Officials, and the Genetic Alliance.

This is my contact information if you have any further questions.

DR. HOWELL: Thank you very much, Michele. I'm sure we have a number of questions and comments. Amy?

DR. BROWER: Thank you for that great presentation. Given the population served by the Maternal and Child Health Bureau but our focus on newborn screening here, where does carrier screening for the moms and dads or prenatal testing fall within the mandate of HRSA or MCH?

DR. LLOYD-PURYEAR: That's certainly within the mandate. We've never had any specific legislation for that, but that generally takes place -- carrier screening is something else, but for prenatal screening, that generally takes place within a clinical setting. We do fund at the state level perinatal screening projects in some states.

DR. BROWER: Yes. I guess just given the definition that it's children at risk for hereditary disorders, if it's a second or a third child, prenatal screening might be indicated based on that. So I was just curious.

DR. LLOYD-PURYEAR: Carrier screening, however, happens by nature of the screen itself for some of the disorders. For sickle cell disease screening or hemoglobinopathy screening in general, you identify carriers with that disorder. That doesn't translate to -- and certainly part of the diagnostic workup therefore

would also include testing of parents, or should include testing of parents. That's also similar for cystic fibrosis screening, that you also identify carriers and as part of that evaluation should be conducting carrier screenings of the parents. But that generally is not a part of newborn screening programs in particular, but it should be part of a diagnostic evaluation.

DR. HOWELL: Denise has a question or a comment, and then Bill I think also.

DR. DOUGHERTY: Thank you, Rod.

One question that I didn't ask Steve Groft but I'll ask you is, is there a plan for an evaluation? These networking things are very exciting. I think they're the most exciting thing happening, and the federal government's support for them. But I think it's important to have evaluations not to show whether MCH did a good job or any of these did a good job but sort of how did the resources available meet up with what people were able to do. People think coordination is cheap, but it's not cheap, and especially when you have all these needs coming up. So I just would ask if you have funds for evaluation of this regional network. That's the first question.
The other one is, is the coordinating center or MCH open to additional suggestions for coordinating center partners?

DR. LLOYD-PURYEAR: First of all, no. With $4 million, we didn't have enough money for an evaluation, although I do recognize the importance of that, and that has been proposed certainly as we continue these, because that is important to show were we able to do what we set out to do, and if so why, and if not why not. Yes, we're open to other partners, and there certainly are other partners that should be part of the National Coordinating Center.

DR. DOUGHERTY: Following up on Amy, I would suggest that ACOG be a partner, and also since the hospitals are so important, the American Hospital Association could be another partner.

DR. HOWELL: There are some other very obvious partners that were not on the list, such as the March of Dimes and so forth. But obviously, that will need additions to the partnering and so forth.
Let me ask a question, and Steve has an answer, but let me ask another question. I'm interested in how -- we've got the coordinating center that you've mentioned, but how are you going to get the groups to get together to talk? Because it looked like the coordinating center is going to draw information more than actually coordinate the groups as a group.

DR. LLOYD-PURYEAR: Do you want Steve to answer the last question first?

DR. HOWELL: Go ahead.

DR. LLOYD-PURYEAR: I bet the answer is no.
Through a variety of telecommunications methodologies, but we actually are having and will have at least once a year a meeting here, bringing all of the principal investigators or project directors for the regional projects. We are participating in all -- these are cooperative agreements, they're not ordinary grants. So it requires a partnership with the Maternal and Child Health Bureau. We also require a willingness to collaborate with the National Coordinating Center. So we are supposed to be part of, in general, not necessarily the day to day activities but the big activities.

But we'll be bringing them together once a year to discuss -- and we're bringing them together in early March this year to actually go back and look at many of the things that they proposed to home in on what's really doable over the next three years and look at ways to partner within the regions, across the regions. The National Coordinating Center will bring resources to that effort.

We'll also have a series of webcasts, videoconferencing. I mean, we proposed a lot. It's not just forming the relationships. It's a communication process that needs to be ongoing and in place. It would be impossible, and also financially impossible, to have the kind of face to face contact that you would want. So we're looking at other methods in lieu of that face to face contact.

DR. HOWELL: I apologize for delaying Steve's response for so long.

DR. GROFT: That's okay. Thank you, Rod.
We will have an evaluation component. We just recently constituted the Data Safety and Monitoring Board. We felt we had to do that first, but we also have a provision for the Scientific Advisory Board, and one of their responsibilities will be an evaluation of the network to see if we've really accomplished what we set out to do when we published the RFA. So we would have some funds from our office to do this, as well as there are set-aside funds within the NIH structure for evaluation studies, and it just requires some planning in advance to do this.
Michele made a very important point, and that is the communications aspect of getting people to work together, and that's a major task of our data and technology coordinating center, that we too are web-based and we have same-time videoconferencing that's available, and we use this extensively in our monthly meetings of the steering committee. Each of the individual consortia can use all of the available resources within the data and technology coordinating center. So there are a number of provisions of how we can work together.
As I mentioned, we hope to join our forces and to link our network with Michele's network and see how we can feed patients both ways just to make things more effective and to make knowledge of each other's activities better known to the entire community.

DR. LLOYD-PURYEAR: Can I just say one thing? I want to emphasize that our initiative really is to address and continue to address the needed service infrastructure, but we view it as a valuable component of the research that's needed, because actually we really do think that a new paradigm needs to be set up that involves newborn screening programs, primary care providers, clinical providers that may not be part of a general research network, and the more traditional researchers in academic centers, especially when you're looking at long-term health outcomes.

So I think it's looking at how to set up those relationships, because they don't work with each other. I mean, they haven't worked with each other in the past. So I think it's getting them to know each other around some specific projects, which will be important over the next few years.
DR. HOWELL: Bill has a question or comment, and then Coleen.

DR. BECKER: Michele, I think you implied or almost stated in your presentation that you expect that some of the results of the Regional Collaboratives could be considered at the level of this area within the purview of this committee. Have you worked out a mechanism?

And I agree with that. I think if you look at the intent of the collaboratives as you've described them, it looks like some of them will achieve best practices or the intent to standardize uniform condition panels across regions, service provision for underserved populations and things. A lot of those obviously are our charge as well. Do you have some thoughts about how you're going to bring those products to this committee so that we can continue to move with them in addition to some of the other things that we're going to be considering?

DR. LLOYD-PURYEAR: Well, that's why I laid out the legislation again for you guys and reminded the committee that one of their charges was to advise on that grant program, so therefore advise on the Regional Collaboratives. We were told to implement the heritable disorders program, and we use the Regional Collaboratives as a methodology to begin implementation of the program. We certainly haven't addressed all aspects of that legislation, so there are other areas for the committee to address, but I think that's a beginning. Certainly we will be bringing, as I'm sure other agencies will, bringing products back to the committee to review.

But we also view the Regional Collaboratives as an opportunity to pilot standards development. If you remember, Mike Watson spoke at either the first or the last committee meeting about the development of ACT sheets and the piloting of those ACT sheets. We'd like to use the Regional Collaboratives to pilot-test those ACT sheets, how well do they work, what are the refinements, take a look at the recommendations for the American College of Medical Genetics' recommendations, or ultimately the

recommendations that come out of this advisory committee and pilot-test them.

DR. BECKER: It sounds like that's going to be an ongoing, active part -- the review of the Regional Collaboratives is going to be an ongoing, active component of this committee's work, and I guess what's left to be devised is the exact mechanism, whether that's going to be a subcommittee type report or continuing, ongoing work. But that's going to be really important to do.

DR. DOUGHERTY: I kind of hesitate to say this, but I'm wondering if the committee would want to consider making a recommendation that HRSA MCHB get additional funds as necessary to evaluate the network. I hesitate because being in a federal agency, I know that additional funds usually means it comes out of some other important program, and I hope that won't go in the minutes. But I really think that this field is new, growing, emerging, that evaluation, a legitimate, objective evaluation of having this coordinating role and information role from regional networks would be really essential. It would be a shame to see time pass by. The best time to construct an evaluation, of course, is before the program, but early in the program is not too late as well.

DR. HOWELL: Dr. Telfair?

DR. TELFAIR: Thank you.

Almost a piggyback on that. I actually had a question in regards to the relationship between the two of what you all are doing, and it's two things. Now I have a second one because of this.

The first one is what will be the structure in terms of the working relationship? Do you see a joint committee? Do you see joint persons, that sort of thing, working together? And maybe as part of that effort, recommendations for how you can begin to look at assessing both efforts. It seems to me that you both are at the same place with that. You're new, you're getting started, and particularly around more and more implementation type of evaluation and looking at your stated objectives.

DR. GROFT: We first started at the staff level that we would get together with the staff of HRSA and our staff in the Office of Rare Diseases to talk about the areas of intersection. We had presented the possibility of having joint presentations at our steering committee meetings, and our group is very amenable to working with the staff, as well as any of the other members, of the network on newborn screening. So I think we've laid the groundwork. Now it's just a matter of implementing how we're going to transfer information, patients, and the knowledge about both programs out to each other.

The network isn't as extensive throughout all of the states. It's unequal distribution, so it's going to need -- there will be the need for considerable information development and dissemination about our network first, and I'm sure about what you have going here from HRSA. So a lot of really groundwork needs to be done to educate everyone about what is available from each program.

DR. LLOYD-PURYEAR: And I think that although the staff recognizes the need to collaborate, the National Coordinating Center has already met with ORD and NICHD. I think it's outlining within the regions. I mean, these really are two different communities. They are researchers within these efforts, but the majority of them are service providers or public health providers. So it's pulling very different communities with very different missions together to look at how they can intersect in a productive way around specific projects. I think the willingness is there, but it's what they would be working together on.

DR. GROFT: Looking at the principal investigators from both networks, there are individuals who have an interest in both areas. I saw some names on the PIs from the newborn screening centers and the networks. So we have some common interests there, and some of our investigators are also interested in newborn screening genetic testing. So I think we have a fertile area to really link up.

DR. HOWELL: I think it's also helpful that you had this tandem presentation today so you each are up to snuff on what you're doing and that will provide a background.
Denise, I'd like to go back. Would you like to make a specific recommendation or a motion concerning the

evaluation issue?

DR. DOUGHERTY: Yes. I'd like to recommend that the committee, this advisory committee, recommend, I'm not sure to whom, that there be funds for evaluation of the heritable disorders program.

DR. HOWELL: Is there a second?

DR. BOYLE: I was just clarifying. It's the regional center program, not the heritable disorders program.

DR. HOWELL: Are you seconding that?

DR. BOYLE: I was just clarifying it.

DR. HOWELL: Denise has made a formal motion. Is there a second for the motion?

DR. BECKER: I'll second that.

DR. HOWELL: Okay, we have Bill.

Can we have discussion about the motion? I think one of the questions is to whom should your recommendation go and from whence would any funds flow should funds be flowing?
Michele?

DR. DOUGHERTY: I guess this committee makes recommendations to the Secretary?

DR. HOWELL: All right.

Any further discussion?

DR. HOWSE: Denise, just a point of clarification. Perhaps we would like to just expand that slightly by recommending that there be a plan for the evaluation, and then the funding would be based on the plan for evaluation, my thought being that presumably when these networks are set up, there's an endpoint in mind and there's a sense of what's to be accomplished. So perhaps as an additional step, staff could provide us with the plan for what would be evaluated that would assist us in building this recommendation for what would be evaluated, how the funds would be used for such an evaluation.

DR. DOUGHERTY: I guess I took that as a given, because you need to have some kind of plan and measures in an RFP for an outside evaluation. But your point is well taken that maybe this committee should be involved in reviewing the plan to the extent possible because of all those RFP issues about not letting things out before they actually hit the street and so forth. Sorry, I'm getting into too much federal red tape.

DR. LLOYD-PURYEAR: I'm just switching hats back to executive secretary. I just need clarification. So there are two parts to the recommendation?

DR. HOWELL: I think it's a single recommendation. I think Jennifer was suggesting, as I understood it, that Denise's recommendation be expanded or clarified, shall we say, to say that a plan be developed. Is that correct?

DR. DOUGHERTY: Well, why don't we say something like that this committee recommends that there be an evaluation of the Regional Collaboratives Program based on a well developed evaluation plan?

DR. HOWELL: And would you accept that, Bill, as a secondary --

DR. BECKER: Yes, definitely.

DR. HOWELL: So we have a motion and a second, and I think Michele probably got that written. Is that correct?
Further discussion?

DR. EDWARDS: I guess I don't understand exactly what would be involved in that, because it's hard to do a $4 million evaluation of a $4 million program. So I'm not quite sure what you're recommending, and that is how much are you thinking about in terms of the recommendation, and are those things that could be done be done under present circumstances? I'm sure you do some informal evaluation already, but I'd just like to hear Michele talk about what she sees is the need for this evaluation.

DR. LLOYD-PURYEAR: Well, within each of the applications we required an evaluation plan to be developed for the grantees themselves to evaluate. There's also an evaluation plan for the National Coordinating Center, but there is not an outside evaluation evaluating the whole concept and process. So Denise is right, that is a gap where you look at success or failure of the regionalization concept. Regionalization is not a new idea and it's not unique to this, and to a large extent I think the need for it is being driven by the rarity of many of the disorders. But we don't have a large evaluation of the whole process.

DR. EDWARDS: And would you address the question of how much you think it would cost to evaluate the program?

DR. LLOYD-PURYEAR: I'm sure it would cost around $500,000 a year, at least.

DR. RINALDO: Michele, I was wondering, it seems to me that that should be part of the work and the purpose of the National Coordinating Center.

DR. LLOYD-PURYEAR: It should be part of the work of the National Coordinating Center to help develop a plan for an evaluation, but an evaluation, a large-scale evaluation would cost more money than the National Coordinating Center has.

DR. HOWELL: Further discussion? We've had a motion and a second. We've had some discussion about the amounts and so forth, and it obviously would be moderately expensive, substantial in relationship to the funding for the program. But is there any further discussion of the motion?

DR. TELFAIR: Yes, I just have a question on that. I understand the motion. I'm not suggesting an amendment to it but just a thought. It seemed to me that there is mutual intent between the two programs, the rare diseases program and what Michele is doing, in the sense that I think I heard you correctly, Dr. Groft, that you also want to make sure that there's a translation effort that goes on to clinical science. I know that in the regional, that's one of the major things that you need to look at too. I was just wondering, maybe for this committee, just a recommendation, that if they're considering recommending and you're setting up a plan, that you don't forget that as part of that process.

DR. HOWELL: Further comments or wisdom about the motion, the discussion, et cetera?
Denise?

DR. DOUGHERTY: I would just point out that, and I think it was referred to before, that the conditions that are being looked at by the ORD do not cover the full range of conditions in the newborn screening programs.

DR. HOWELL: We have a motion and a second and a considerable discussion. Are folks interested in voting on the motion? I see heads nodding and so forth.
Those favoring the motion, raise your hands.

(Show of hands.)

DR. HOWELL: Those opposed to the motion?

(No response.)

DR. HOWELL: Those abstaining?
(No response.)

DR. HOWELL: So was that unanimous? Did you raise your hand, Jennifer?

DR. HOWSE: I don't think I'm a voting member.

DR. HOWELL: Oh, you're not. Thank you very much. I'm glad that you're so careful about those things.

Madam Secretary, please reflect that it was unanimous.
Is there any further discussion?

(No response.)

DR. HOWELL: We'll leave for lunch a few minutes early. You'll have to fight with everybody who are building all these stands outside for the inauguration, so a few minutes will help, and we'll resume at 1 o'clock and hear some exciting things about follow-ups that are happening elsewhere. Thanks very much.

(Whereupon, at 12:52 p.m., the meeting was recessed for lunch, to reconvene at 1:00 p.m.)

AFTERNOON SESSION (1:06 p.m.)

DR. HOWELL: We're coming in, and Steve is going to be here presently. Thank you very much for getting back promptly, the committee. We're going to continue on the same theme of some infrastructure follow-up efforts, and I'm pleased that Judy Tuerck, who has been involved in newborn screening and treatment for many years, is here to discuss the CDC database for the long-term follow-up of infants identified by tandem mass spectroscopy.

Judy? I think you probably are going to need to stay seated and turn on the microphone, please. It's the green button in the center. Excellent.

MS. TUERCK: First of all, I would like to thank Michele and Dr. Howell and Coleen Boyle for making it possible for me to be here to share this information with the committee.

I've had sort of a peripatetic background. I've spent 26 years working as the nurse in the metabolic clinic at Oregon Health Sciences University, and also about that long doing newborn screening follow-up for the Oregon State Regional Program, and at the same time doing practitioner and parent education around newborn screening. So for me, this project has been a labor of love because for all this time, I've recognized the need and the importance of long-term follow-up, but unfortunately short-term follow-up has just taken up so much of my time and so much of our energies over the last 25 years that it's always hard to get on to long-term follow-up. So this opportunity to address this problem in a real formal way, through the CDC, has been to me just a wonderful project to work on.

I'm here really as a representative of the group of folks who have been doing this. We have had a cooperative agreement with the CDC, and the grants were given to Oregon and to Iowa, and we included Idaho because they're part of our regional screening program, and they were also going to be starting tandem mass at that time. Coleen Boyle at the CDC and Aileen Kenneson and Katie Kolor have been very, very helpful to us, sort of steering us through the amazing complexities of this project.
In Oregon, myself and Sara Copeland, who actually is the metabolic doc now in Iowa, which has been terrific because now I don't have to pay her off my grant but she can be paid off the Iowa grant, and then Christiane Winter, who is our programmer, and Lori Paradise and Cary Harding in our metabolic program there. Sara, as you can see, is now in Iowa, and the team of folks in Iowa who have been working on this, and the folks in Idaho have also been working on this grant.

Just to remind folks that long-term follow-up is something different than short-term follow-up. Short-term follow-up identifies the children and gets them to the place of diagnosis and confirming that indeed they have a disease or they do not have a disease based on screening test results and confirmatory test results. Long-term follow-up kicks in once the child has been diagnosed and is on treatment, and in my perfect world which I know doesn't exist, it would continue really throughout the life of the individual regardless of where that child is being treated in this country.

We have been telling folks for at least the last 15 years that they need to be doing long-term follow-up, and it is in every single one of our guideline papers that we published. Unfortunately, none of us are doing a very good job of it. The kinds of data that we need to be collecting you all know, as well as I do, that there's a whole list of things that are not on this slide but that could easily be put on this slide. These are just sort of the seven or eight most important things that we think about in terms of long-term follow-up.

For me, it has been horribly frustrating because I know that those data are there. They're sitting in the kids' charts, they're sitting in treatment centers around this country, and we just don't have a way of getting the data out. The problems with long-term follow-up, at least from my perspective, both as a clinic nurse and in the screening program, has been that it's very, very frustrating and expensive to do long-term follow-up because you've got to collect a lot of data, it's very expensive to collect it in a uniform and comprehensive way, and if you get inconsistent data collection between centers, then it's difficult to compare the results.

I think one of the limitations to long-term follow-up has been that none of us really have known what data we need to collect. We sort of think we know what data should be collected, but when you actually sit down and begin to make up the list, it becomes complicated pretty quickly. The other problem in the United States is that any given program in this country is going to have so few patients that we'll never get a big enough N to make data analysis worthwhile. None of us, as I've said, are doing a good job with long-term follow-up, as Brad said this morning, and that's been true for 40 years now that we've been screening for these diseases. None of us are doing a good job of the long-term follow-up. We're relying on what's coming out in the literature from individual docs who get around to writing up the case reports or who get around to writing up surveys. I'd propose that that just isn't good enough anymore.

So the question is why does it matter? If we've been not doing this for 40 years, why all of a sudden should we be doing it? I would like to use as my example of why we should be doing this one rare disease, which is galactosemia. For those of you not in metabolism, this disease causes a neonatal emergency in that about 80 percent of the kids will develop nasty liver disease, and about a third of them will die in the first two weeks of life of overwhelming sepsis. Way back in the '60s, this disease was recognized. We also knew that if we took the kids off of lactose and we put them onto soy-based formula, that within 24 hours of doing that the liver disease resolves, the sepsis sort of settles down and goes away, and they absolutely look like roses and they're absolutely fantastic.

So, wow, what a great disease to screen for. So all the states -- I shouldn't say all the states. Many states rushed to add galactosemia to their screening battery, and it was really felt that all you needed to do was to put the kids onto a soy-based diet and things would be just great. Actually, I wanted to say that universal screening for galactosemia was finally achieved last year, 40 years after we started. So the idea that we're being slow in implementing tandem mass spec to me is sort of laughable, because it's very, very fast in my experience with screening.

The disease is rare. We've had trouble getting children to come into clinic because their docs don't want to refer them because they think a milk-free diet is so easy to do that we don't really need to worry about this too much. It wasn't until 17 years after screening for this disease was implemented that the first papers began to be published about problems in galactosemia. Those papers continued to be published throughout the '80s and into the early '90s.

I have included this because this was a little comment, an anonymous comment that came out in the Lancet in 1982, which is now 20 years ago, 20-some-odd years ago. We now have 10 years to go before this prophecy comes true. We still have not done any kind of comprehensive survey. The only thing which

has been done, and again thanks to HRSA, who was able to sort of give Neil Buist and Diane Waggoner I think it was about $10,000 or $15,000 to do this worldwide survey. This is the only study that has been done in this country -- it's been replicated in other countries like Germany -- that tells us the breadth and depth of the kinds of problems kids with galactosemia get into.

We are now at 40 years out from the beginning of galactosemia screening. We still don't understand the basic problem in galactosemia, nor do we have appropriate treatments which will counteract the side effects. Should we be screening for it? I think absolutely, because we're saving lives. There are a number of diseases that we're now screening for which save the children's lives but do not necessarily prevent the complications. The important reason for trying to understand this is that parents have a right to know, if the complications cannot be prevented, what screening is actually doing for their child.

We are now in a position in this country where we're now recommending, instead of one disease, we're recommending that we start screening for 30 rare diseases. I think what's going to happen, and I already see this happening in our own program, is that some of the MCAD kids will be seen to be so easy to manage and they will be seen to be doing so well that long-term follow-up really isn't necessary or important for those children. There will be benign conditions like SCAD and 3MCC, which are touted to be benign, but are they really? We have no idea if they really are benign for 100 percent of the affected kids.

I do think we are in a position in this country right now that we know we need to be doing long-term follow-up, we've been advocating it for 40 years, but we're in a unique position now to begin actually doing this. The problem that we have had is that we don't have a tool, and I think we need a system to pool the data and to share it both nationally and internationally, which brings us really to our little project, which was the development of the tool.

Our contract with the CDC was that we needed to have MS/MS implemented in our programs, which we did, and really in the beginning our contract with them was to develop a paper and pencil long-term follow-up protocol. So what kinds of data should be collected, and how do you collect them? And then do it the old-fashioned way.
What happened, of course, is that after we had done the literature review and we had organized our data into protocols, the protocols for each disease were 26 pages long, and I about had a heart attack. I just looked at that and said I'm not filling that out. And if I wouldn't fill it out, and I was one of the people that wrote it, I knew that there was no way we were going to get this data out of treatment centers. So we knew we had to put it into a computerized database of some sort.

We also developed fact sheets that other folks have gone ahead and done the same thing, but we also have done that because we had just done this amazing literature review and I just hate to have all that information go to waste.

When we started looking at the possibility of putting this onto a database, we knew that we needed something that was going to be HIPAA compliant, and needed something that was going to be reasonably portable, and that it needed to really be adaptable so that it could talk to other information systems. On my list was this automatic data collection. The less time that you have to spend abstracting these data, the cheaper it is and the more likely it is that you're going to get the data. We're now in an information age where a lot of this stuff is computerized. Why can't our computers do our abstracting for us?

I think we may have an answer to that. We hired a database programming who was actually a fish biologist, for heaven's sakes. I mean, I can't believe this lady has done the miracle that she's done. She's developed this database for us, and the important thing on this slide is that it is an adaptable database so that we could add 500 metabolic diseases to this thing if we wanted to. We can also use the framework of it to include diseases like CAH or thyroid disease or sickle cell or whatever needs to be done.

We really designed this to be used at the point of care. So in some states, for example in Oregon and Iowa, this is at the university metabolic program clinics. In other states that don't have metabolic centers, this is being done in the state health division clinics who run metabolic clinics and they bring in outside

specialists to take care of the kids.

I wanted to share with you a little bit about the kinds of cases that we have found. In the beginning, based on what was available in the literature at the time, we expected to find three or four kids in Iowa, three or four kids in Oregon, maybe one or two kids in Idaho. So far, we have had five times that many children identified with real disease. This does not include PKU or any of the other metabolic diseases. This is just the new MS/MS diseases.

The other thing that's important on this slide is that in Oregon five of these kids have been identified on the routine second test. So not all of these diseases are going to be identified in the sample taken in the first day or so of life, and I was very distressed to hear this morning that Connecticut has traded tandem mass spec for their second test, which we had considered doing as well because we had been led to believe that tandem mass is so specific and so sensitive that all of these diseases will be identified in a sample that's taken in the first 24 to 48 hours of life, and we are not finding that to be the case.

The kinds of kids that we're finding on the second test are carnitine transport disorders, CPT1, arginase deficiency, homocystinuria, and one child with VLCAD. We've also had some additional cases that we identified on the second test, but those are really cut-off issues with the cut-off levels that we had and I don't really feel that they were true biologic variants. But these cases we really consider to be biologic variants who are less likely to be picked up on the first test.

The incidence of these diseases in our states has been very interesting in that here we are in Idaho and Alaska with an incidence of somewhere between 1 and 2 in 3,000, and look at Hawaii, who allegedly has all these kids with MSUD and organic acidemias. We have yet to find one patient in Hawaii, until I go home, and there will be two or three probably. It's been an amazing, eye-opening experience to do this.

The way that we've been able to use the database just very preliminarily in our cases, because we're still at a stage where we can do it quicker by hand than we can playing with the database, but we've had 37 cases identified in Oregon and Idaho. Thirteen percent of those kids have been symptomatic at or before diagnosis. We've had three deaths, which we're not used to seeing deaths in newborn screening programs. So that's been traumatic for all of us, as well. But on the up side, 92 percent of the kids are "doing well." Mind you, these children are only just turning 2 this October, or last October. So they're all just now coming into their second year.
We can also track things like hospital visits and that sort of thing with this database.

You all have copies of these screen prints in your packet. So I'm not going to spend a lot of time, but I wanted to just show you how the database is organized and how we believe that it can be used. You can see patient intake information, laboratory testing, metabolic episode issues, and then office visits and follow-up status.
Christiane has organized the large buttons on the left-hand side. They're the buttons you saw on the initial screen, and then across the top are drop-down boxes and additional boxes which pertain to that big button on the left. So intake information along the top here are the patient demographics -- caregivers, pregnancy history, family history, and diagnoses. These are all meant to be one-time data entry things. You're not going to have to change this very much. I'm going to run through these very, very quickly.

We have drop-down boxes to make life a little bit easier for people. Pregnancy history, we were interested in collecting information on liver disease in the moms and any other prenatal problems that the mothers may have experienced. Family history, the drop-down boxes -- and again, these are in your packets -- primary diagnosis. If you can see here that under the disorder drop-down box, you can add any metabolic disorder you wanted to add.

Newborn screening test results. Again, I would love to be able to see us import this data from Brad's shop, for example. Why can't we get it from the state health division directly? Why do we have to reenter it? Here's the analyte list, and again you can add whichever diseases you're screening for. Metabolic studies. These would be either confirmatory studies or they would be ongoing management studies. Again, the drop-down list of the various tests that can be ordered and the laboratory that's doing those

tests. Amino acid studies and mutation analysis. Then basic laboratory studies like UAs and chemistries and blood counts and things like that are also in the database.

We're very interested in trying to collect information on the kinds of metabolic episodes that these kids may have, so we have tried to build this in to count these sorts of things. The question came up very quickly that sometimes what might be considered a metabolic emergency by the family is not necessarily a metabolic emergency but might represent the anxiety that the family is feeling in the early days before they realize how stable their child might be, so we sort of say "worried parent," which is not inconsequential. It is a serious problem that we have to work with the families to sort of help the families cope with these diseases.

This has stopped working. It's not allowing me to enter. It's now allowing me to advance. Thank you. Signs and symptoms of metabolic episodes, and then metabolic management visits. Again, this could be tailored to be an endocrine visit or a hematologic visit.
One thing that I wanted to point out to you is the ICD9 and the CPT codes. I wish it had been my idea. I would be so proud if it had been, but unfortunately it was not. It was Pat Lawler at the CDC that suggested that we add those, because that will allow us to get back into the billing offices and to pull out the charges that are actually made for the kids on the visit A, B, or C, and include all the labs and medical foods and things like that. So we're pretty excited about having that on our database. Then this is just sort of trying to record problems that the child may have had between visits.

Most of us do informal developmental evaluation on the kids all the time when we see them in the clinic, and we wanted to be able to at least know whether or not the physicians or any of the team members had concerns about the kids at their visits.

The primary care provider, medical summaries, some indications about therapy, and what is their therapy. This database is not meant to prescribe therapy. It's really meant to just record what it is. Again, this area we believe could be expanded by somebody who wanted to do a clinical study on MCAD disease efficacy. For example, they might want and need to have much more detailed information about the dietary fat intake for these kids, for example, and this database would be easy to adapt to collect that kind of information.

Formal developmental evaluations, specialist office visits, again with ICD9 and CPT codes, and then the last slide is sort of where are we in terms of is the child still active in the clinic, moved away, dead, whatever. We've also developed data collection sheets, which you have in your packets, which are meant to be used by a physician or by a team member in clinic so that, number one, they remember to collect all the data we want them to collect, and they can use them to guide them through the visit as well.

We really think that this database, which is still pretty rough -- I termed it as a nice little Hugo. It's a little car that runs, it does pretty well, but it's not a Cadillac, it's not a BMW yet. But it's the beginning of something we've never had before. We think we can use it to evaluate the efficacy of screening, and I think also that, with a little playing and a little updating, it could be used for clinical studies by folks at the NIH who are trying to understand these diseases.

In my perfect world, my utopia would be that instead of having this done in every state health division, that we would have this database or something similar to it in every single clinic in this country. Wouldn't that be a story? I know that that's not a real possibility, but if we could just start with the newborn screening diseases and then gradually add additional diseases. We were talking this morning about how they're screening for Krabbe's disease in New York. Well, a lot of the lysosomal storage diseases have great treatments now, but we don't have a screening test. So we're always sort of playing catch up with these things.

But we could at least begin to collect data on natural history of lysosomal storage diseases, although that's pretty well known, perhaps more so than some of these organic acidemias and fatty acid oxidation disorders. I think it would give us the possibility of collecting information on the benefits and the potential complications of these diseases before 20 years has passed, which has been traditionally what happens.

You don't know what kinds of problems the kids are going to have until 20 years down the road, and I think it might give us a heads-up on that.

We need to do a bit more work on the database. We need to be able to Web-enable it, and we need to add at least the other metabolic disorders, and this would be quite easy and simple to do. We would like to investigate the ability to collect automatic data from the billing offices and from the laboratories. There's a new program out which Jim Hanson had talked to Sara Copeland about which is something called a parser program. I'd never heard about it before, but it's a software program that you program to read electronic medical records, and most of us are going to electronic medical records now. This program goes in and it pulls out automatically the data that an abstracter would normally pull out and theoretically could eliminate the need for abstracting for a lot of these data points. So we're very interested and excited about that.
I think if we tried to model a national metabolic center data repository after the CF and the national cancer databases, they've done this before, they know how to do this, but it certainly has never been done for metabolic disease.

As far as ongoing research with this particular database, we would really like to see how easy it is to implement it in other states, and certainly to evaluate the costs of collecting the data and to evaluate the interactivity of the database. How do we pool this data? How do we analyze it? Obviously, the folks at the NIH have got a lot of experience in doing this, and to evaluate the database to see how functional it will be with other programs.

I think some of the considerations are obviously confidentiality and informed consent. I don't think I would want to do this again unless we go to an informed consent process for the next go-around, which would be relatively easy to do. We're very interested to know how easy it would be to get this database out into metabolic centers in individual states and then feed that data either back to the state or back to a national data pool who could then feed it back out to the states. But I think that something like that is really a very doable thing.

Obviously, we would really like to continue this project if funding were available for a longer period of time. We've already collected data. By the end of this year we will already have collected data on children for three consecutive years who have been identified in our program. Additional funding would allow us to continue to collect data on those same children plus any others that were born. We would have the oldest ones in school. So we would have them out to five or six years of age.
We've had such interesting results in the northwest region in terms of the kinds of diseases that we're finding and the number of diseases that we're finding on second tests, and we have lots of states who are interested. I think that there are a lot of other places in the country that would be very interested in trying to work with this database to help us see what we could do.

I will stop there and let you ask questions.

DR. HOWELL: Thank you very much.

From your comments, I gather that at the current time you do not have informed consent.

MS. TUERCK: We do not. We were able to get an IRB waiver to informed consent at OHSU because we were going to be doing chart reviews and they felt that the chance of doing any harm would be low. But I'm feeling constrained by that, and you can't ask parents any questions about it.

DR. HOWELL: And you could not pull out the patients for research or recontact or anything of that nature.

MS. TUERCK: Right.

DR. HOWELL: Because you lack that.

MS. TUERCK: Right.

DR. HOWELL: I want to go back to the second test issue, because that's a very important thing, because most states in the country do not do a second test.

MS. TUERCK: That's correct.

DR. HOWELL: I'm surprised and somewhat alarmed by the second test results, and I have a question. One is, have you had an opportunity to go back to the original test --

MS. TUERCK: Oh, yes.

DR. HOWELL: -- and be very certain beyond a shadow of a doubt that the first test was negative?

MS. TUERCK: I believe that they have sent, in each of those cases, those cards have been retested not only in the Oregon lab but they've also been sent in some cases to outside labs, like Baylor or Mayo or to Dr. Millington to assay them. Harry could probably answer the stability question. We do our second testing at around two weeks of age. We certainly know that the fatty acids, for example, are not necessarily stable over a long period of time, either in the child or in the blood spots themselves.

But yes, we've tried to do that.

DR. HOWELL: And the outcome when you've actually sent the spots to additional places, they verified, using their cutoffs and their technology, that they were negative also?

MS. TUERCK: Yes.

DR. HOWELL: That's a very important thing to continue to do.

Piero has a burning question, I think on the same issue. I can feel the burning coming from over there. (Laughter.)

DR. HOWELL: But what data do you have about any persons again when you show such divergent data from Hawaii? My first question was do you have any data on patients that might have been missed?

MS. TUERCK: We know we missed one baby with LCHAD, not in Hawaii. In Hawaii we had one baby who was identified prior to their joining, before they started tandem mass officially with us, that we identified with citrullinemia, but that baby had already been clinically suspected.

DR. HOWELL: So during the time that you had an incidence of 1 in 15,000, you're not aware of any persons that have been missed?

MS. TUERCK: No. We had two babies in Hawaii with intermittent MSUD who were identified during that period of time, but their test results were normal, and that's well known for intermittent MSUD, that you wouldn't necessarily find those kids on an early sample.

DR. HOWELL: So the MSUD patients were indeed identified outside the screening program in Hawaii during that period of time.

MS. TUERCK: Yes, and we've had one or two in Oregon over the last 30 or 40 years as well.

DR. RINALDO: My question really continues on the topic, because I really think you're making a very strong statement about the need for a second test. Obviously, that is a conclusion that your program may reach, but there is a need to clearly document the analytical performance in the analysis of the first one, because if you say that the second one is necessary because the first one, for a variety of reasons that might include perhaps less than adequate cutoffs, we need to do it twice because we're not sure about the way we do it the first time, I don't think that's really an acceptable argument.

MS. TUERCK: Well, it's not a matter of we're not sure it was okay the first time. We believe that the results are correct the first time. They just aren't abnormal. I don't care how much your instrument costs, you can't detect an abnormality before it develops.

DR. RINALDO: I want to go on record that I emphatically disagree with that statement and I really think you have to come up with some hard evidence, because if you set a cutoff very high, then the abnormality be evaluation and elsewhere might be there. You just don't want to see it. So I really challenge, if you'll allow me, that statement, because if you show me the data are coming from Millington or Baylor or us and say, okay, this is what we got, this is what they got, and show a reasonable agreement in the conclusion on the analysis of the first pass, if there is an agreement, then I will say okay, there is a point here made to have a second test. But without that evidence, I really don't think you can make that statement.

MS. TUERCK: Well, obviously I'm not a biochemist, but I know that the lab has had the original specimens tested not just by our lab but by other labs and that the results were normal. That's all I can tell you at this point. Our cutoffs are -- as everyone is struggling with cutoffs, we too are struggling with cutoffs, but I think that there will be a subset of children with these diseases who will not necessarily be abnormal in the first day or so of life, and we've known that's been the case for homocystinuria for 25 years, and we know now that we also find about 10 percent of our children with thyroid disease on the second test, and we find about 5 percent of the CAH babies on the second test, not just in our program but in Texas and in other places where a second test has been used.

This is not necessarily news. It is certainly news for tandem mass diseases. We didn't really know what we would find and we were interested to know what we would find, and we are shocked by what we're finding.

DR. RINALDO: Just one final point. I really believe that these data should be really made available for peer review. It really is extremely important, because statements without evidence to back them up, I think they can have unintended consequences.

MS. TUERCK: Yes, the paper is in process.

DR. HOWELL: Derek?

MR. ROBERTSON: Just two things. One is that I'm pleased to see the work on the long-term follow-up, because I've said all along that the long-term follow-up is probably just as important as the initial screening process. But could you just explain for me and for other lay people why you'd be so confident that the first test would be always correct?

DR. RINALDO: Nothing is always or never. Always and never don't exist in the kind of work we do. My point is this, that if a patient with a certain disease has a value of, say, 100, whatever unit you use, the real issue is the cutoff. One lab may have a cutoff at 80, another lab may have a cutoff at 150. So if you use the cutoff of 150 and you call it negative but another lab would pick it up, you have a moral/ethical duty to challenge that cutoff and how it was determined, because chances are the cutoff was set up wrong, or it could be a statistical fluke, a million reasons, a variation in the method.

Again, the whole point I'm making is just make sure that the concept of negative here is a subjective concept and not an objective concept. That's really the difference.
MR. ROBERTSON: But does the cutoff, again as a layperson, does that cutoff change potentially as the child gets older? In other words, it would be different at two weeks than it is at two days?

DR. RINALDO: In some part. The point is even for the same age range, changes from lab to lab, from program to program. So when we talk about uniformity standardization, this is really one of the fundamental issues in finding the right balance between excessive noise and false-negative results. The story of tyrosine is the best example. At one point the program in North Carolina got to a point where the cutoff was set at 1,000 micromolar because they were sick and tired of having a lot of false positives. But

guess what? At that level, you will never diagnose a case of tyrosinemia type I. It's just incredibly high. So it becomes an exercise in futility to test for tyrosine when you have a cutoff that is so high that it will never be exceeded.

MS. TUERCK: I would totally agree with you, Dr. Rinaldo, because I don't believe any of us are screening for tyrosinemia at the moment. We say we are. Everybody's got it on their list, but if the proper cutoff for that would be less than 150, most of us have cutoffs around 500. So it makes it very unlikely that we're going to be able to find a real tyrosemic on first or second screen with those sort of problems.

DR. RINALDO: Unless you use a second-tier test.

MS. TUERCK: Exactly. We need succinylacetone. Absolutely.

DR. HOWELL: The tyrosine is probably one of the best known situations, because particularly small gestational preemies have a dramatic elevation of tyrosine that can persist for some time, so there's frustration with looking for high levels. But I think the key thing is you really need to screen, and when you get these positives, to then consider a second-tier system like is used in Canada routinely with succinylacetone. So I think the key thing is you've got to be sure you don't miss patients whenever possible.

But I want to be sure we continue the second screen thing, because if one recommends that all patients in the country get a second test, that's a big-time recommendation. How many states currently do a second screen?

DR. THERRELL: Eight states mandate a second screen on every baby. Three states recommend it to the point that they get greater than 80 percent compliance. In those states that mandate it, they have data supporting the mandate primarily with hypothyroidism and CAH, and an occasional galactose, an occasional something else.

DR. HOWELL: Thank you very much.

Steve, you had the light on for yourself before.

DR. EDWARDS: I wanted to ask you a question just about technically how you do that. Obviously, you've got the captured population in the hospital for the first test, so that's pretty easy. But how do you get that second test done, and what sort of compliance do you have with getting the second test done?

MS. TUERCK: Well, in Oregon we were one of the original Guthrie sites, and Bob in those days was worried that PKU might be missed if the sample were taken early. By early then, that was three or four days of age. So we've always had a two-test system in Oregon. We have looked at dropping it several times. One was right around the time we started thyroid screening, and the second time was just in the last couple of years, starting tandem mass, and we decided that we really wanted to wait and see if there were any diseases that we find on the second test.

But our physicians in the community do a second test on the babies. The kits are -- there are two cards joined together that have the same identifying number. Those are torn apart by the hospital. One is given to the parent, along with the envelope to put it in and to keep it safe, and they take it to their physician's office on the first visit, and he or she collects the sample at that time. We have a very good compliance rate with the second test. I think over 95 percent of the kids in Oregon are tested twice, and most of the time that second card is retained by the family.

We did a study on this several years ago and about 3 percent of the samples on the second test came in on an unmatched card, meaning that the doc had to use a different kind of card because the parent had lost or destroyed it. But most of the time they're pretty careful about them.

DR. HAWKINS: Coming back to the issue -- I talked with Piero about this, but to find out that there's not a

uniform standard on these tests from state to state, from lab to lab, to me that's one of the most troubling things I think I've heard, knowing that any type of testing is going to be consistent from one state to the next. Is that true with just about every test that goes through this? I mean, to me it seems like we're talking putting the cart before the horse in some of the things we discuss here. If we say there's not a uniform standard from lab to lab, from state to state, it sounds like we need to step back, set some standards before we move forward to do some of the things we really want to do.

DR. RINALDO: Well, I'm glad you say that because that's exactly one of the goals of the Regional Collaboratives. Whenever we say, okay, this state does MS/MS, this state does MS/MS, and so on, it will be an extremely optimistic assumption that the performance, the analytical performance is comparable. It's not. This is one of the elephants in the room that nobody wants to talk about.

DR. HAWKINS: So you're telling me that a lab, say, in Oregon or in Mississippi, there's maybe just one person that comes up with a set standard? I mean, they set the standard for the lab? They decide what the cutoffs are? Do they not have literature to go to, to give them good guidance or call you or call somebody else?

MS. TUERCK: When we started, unfortunately, the private labs have not been willing to share their cutoff data with folks. So the states who have started this have had to rely on data that's already been generated by other state programs doing it. So when we started in 2002, we took the cutoffs from all of the states that were currently doing tandem mass and we looked at the cutoffs, we looked at the analytes, we decided sort of as a group with the advice of our advisory committee that we didn't want to screen for non-treatable diseases. So things like glycine and some of the non-treatable markers we decided not to screen for and winnow those out.

But you set standards by doing your own validation studies in the laboratory so that you coordinate your machines and you know that the answer you're getting is reasonably correct. But we set our cutoffs based on those validation studies and what other states were doing, and the other states have been wonderfully helpful in sharing those sorts of data. We need to do more of that on a national basis because you're exactly right.

DR. HOWELL: Bill has been waiting down there patiently.

DR. BECKER: Thanks. Yes, Greg, Piero is exactly right, and just to expand on Judy's comments, this is a learning process. This is the way methodologies are implemented in any laboratory test, whether it's a clinical laboratory test or a reference laboratory test or a public health laboratory test. These assays are evolving. We are learning about the assays and their performance all the time by gathering data, particularly long-term data, and there's a tremendous need for this evolution to continue. These tests need to mature with increasing standardization.

The comment that there are different reference ranges probably between any two hospitals in your city for many of the analytes that you routinely test for, and that's not the basis on which to make a decision. It is standardization, the assurance of quality, the maturing and evolving of standardization efforts, all of those activities are critically important for us to continue to improve these programs like they need to be improved.

DR. HOWELL: Amy?

DR. BROWER: This is a different topic. It's more on the databases. So do you want to finish this topic first?

DR. RINALDO: Yes. Because you brought it up, is it a fair statement that the cutoff used at that time was 10 times higher than what in general is used by other laboratories?

MS. TUERCK: I'm sorry. For what?

DR. RINALDO: For the LCHAD case that you mentioned earlier.

MS. TUERCK: The LCHAD case, I can't comment on that. I would defer to Mike Skeels and to the metabolic folks. My understanding was that it was -- I don't know where the cutoff was in relation to other cutoffs at that point.

DR. RINALDO: But do you have any knowledge that the cutoff was reduced after?

MS. TUERCK: Yes, yes. But that was for a missed LCHAD. The rest of our cutoffs are comparable with what other states are doing, and they're constantly being reevaluated as more babies are tested. We made a change in one of the cutoffs last week, for example.

DR. HOWELL: A very excellent presentation about long-term follow-up.

DR. BECKER: Amy had a question.

DR. HOWELL: I will come back to Amy. I haven't forgotten her.

But the thing is that she's brought up the very critical issue, and there are two things that are very heavy on the table that we've got to keep on the table. One is, is a second test really indicated or necessary? That's a very big-time decision, and that would need to be made with as much data as could be brought to the table. The other thing that has been around the table a lot is cutoffs and how they're determined, and the methodology and technology surrounding newborn screening, which is a very big issue.

Amy has some important questions about data.

DR. BROWER: Yes. Thanks, Judith, for your presentation. I commend you guys for taking the big leap into trying to integrate all the data, and I would just encourage you to look to NIH databases and database practices to really use best practice, because you really want to include data dictionaries and maybe publish them so other people can follow your lead, data normalization, because it's not only the data capture and storage. We really want to integrate it and be able to analyze it. From what I saw today, there's still a lot of narrative text built into your database, and that's not queryable.

So we can capture that narrative text, and it might be useful clinically or anecdotally, but we can't do any data analysis on it. So I was just curious, one question about what platform you developed. I know that there are parsers and things that NIH and other groups that can assist you in not only reading e-medical records but also incorporating medical literature and other things that you want to integrate into one database.

MS. TUERCK: You're absolutely right. We put this on -- I think it's on an Excel base. I'm not a computer person. Everybody says, well, if Judy can do it, everybody can do it. I recognize that it's rough. We tried to keep the text information down to the least amount possible, but as yet we really don't know what kind of data we're going to be running into. As we begin to accumulate these children and are able to analyze their data to see what kinds of complications or problems they're running into, then we can build that in.

As I said earlier, it's a nice little Hugo, but it's not a Cadillac or a BMW, which is what -- my plea to this committee is to recommend that this be used as a starting spot and to be able to move forward with it.

DR. BROWER: I think it's a great learning spot, but you might learn from what you've encountered. Maybe at the first glance we can't get all the data, but let's get 80 percent in a database that we can query using best practices, Java, Oracle, all those different informatics tools where we can really integrate the data in real time.
MS. TUERCK: Exactly.

DR. TELFAIR: It looks like you developed this using Access. Was it also on the SQL server as well?

MS. TUERCK: I don't think so, but then I'm not the expert in computers. We had to leave our home.

DR. TELFAIR: Oh, okay.

The other question I had is that when you're looking at the database, you have a lot of fields that are there. Have you begun the discussion of a minimum data set? Because if you're going to do the cross-sharing of that, not everybody is going to be looking at the same set of variables, but it seems to me there's a common set of variables you would look at as well. I mean, I compliment you because I agree that it's a real good start and I think it's critical. Just at the level of application, you're looking at what would be a minimum data set to work from.

MS. TUERCK: No, we haven't done that yet. Trust me, I think we're on version 8.2 or something. I mean, we've talked about this forever. We've made it a bit more inclusive than we know it needs to be, because we're not sure what data needs to be collected exactly. So I think once we get a critical mass of kids into that database and are able to actually start running reports and looking at it -- if, for example, thyroid people wanted to track those diseases, we would obviously need to strip out all the metabolic language and they might have different languages and different data points that would be put in.

But I think that all the family history stuff and pregnancy and all of that, mutation analysis, those things would be common to virtually all the diseases and you wouldn't need to add that. So those would be kind of part of the beginning of a minimum data set.

DR. HOWELL: Bill?

DR. BECKER: Judy, two questions. One, what is the status of the database right now? I think you implied it's inactive or running out of funding or something. The second is kind of a thought question. As I was trying to think about how a database like this could be utilized on a wide scale, I'm reminded of, first of all, I would like to think we would be able to produce a recommendation that wouldn't duplicate something that might already be out there. In other words, who does a lot of database entry for public health programs from the clinical setting right now?

The thing that came to my mind is the cancer registry. There are cancer registrars. That's actually, as I understand it, a certification process to be a certified cancer registrar, and it's a national database. All states are required to report into this national database. It's uniform. It's about 30 fields, if I recall. It's been a while since I've looked at exactly the data type entry.

It's the kind of thing that, as I understand it, it's mostly in the medical transcription or medical records departments is where these registrars tend to operate out of. But I'm wondering what your thoughts would be on something like a heritable disorders registrar or registry that would certify people, even though it's pretty easy to use. Certification I think is important for other reasons, but what your thoughts on something like that would be.

MS. TUERCK: That's about a fifth generation question. We're not even close to that. To me, there are lots of ditches to leap in the meantime. I think that the informed consent process is going to be critical because there's already been an issue of if you register all the kids with metabolic diseases, that somehow the state is keeping track of all the kids with mental retardation, and that is not what this is about. I think that it's down the road a ways, if something like this would fly.

We're in the last year of a three-year cycle. So what worries me about this database is that if no further funding were allocated to it, it will probably die. We don't have it hooked up in any official way yet to communicate our results, even back to the state health division in Oregon, but we can get it there. But it will take some additional time and finagling. But I would love to see it move forward. But I agree that it would be wonderful to have a national repository on heritable diseases, absolutely.
DR. HOWELL: Coleen has some comments.

DR. BOYLE: I guess I wanted to follow up, Judy, on the issue of informed consent. I've been musing

about it since you put it up on your slide. Most of our public health systems that have surveillance programs, they operate under the auspices of public health, it doesn't require consent. The reason is that it is sort of a public health mandate and we are assuring the public's health. Then issues of consent always minimize who participates in that program or in that follow-up.

So I guess there's some utility in getting consent. Maybe it allows access, it allows special studies to occur, but it also diminishes from my perspective perhaps the utility of the program a little bit. So I don't know, I would have to think in more detail about the issue of informed consent. I think it's very complex.

MS. TUERCK: It is.

DR. BOYLE: This was viewed more as a surveillance and tracking programs. Cancer registries don't get informed consent, and they do linkage in terms of treatment-related aspects. When we get the consent part, we really limit who gets involved in that linkage and follow-up.

MS. TUERCK: If you think of this database being deployed in a point of care clinic, the problem we've run into, for example, is that it's difficult for us to get the birth records on an affected child because those may not be in our medical records. If the child dies, we don't have access to the death certificate. Those sorts of things are limiting. We're finding ourselves limited by working under a waiver.

DR. BOYLE: I actually think we're going to hear a little bit more about this from our next speaker. But I think all of it should be within the purview of public health. Those are all essentially national databases in many ways. So in order to assure tracking of children long term across state boundaries or state lines, we need to have this capacity in there.

MS. TUERCK: And I think it's important for us to use those people as models, because this ground has already been plowed. We don't have to do it again. But I think in order for us to be able to capture this data, we're going to need to be able to capture specifics on development and general health that we may not be able to do under a blanket thing. But again, there are different models, and one could try it with informed consent in one center and with a waiver in another center, for example, and see how it goes.

DR. HOWELL: Steve?

DR. EDWARDS: I commend you for the very important step of long-term follow-up, which I think is absolutely critical. But help me with one thing. You know, computers are wonderful, but some of us came along in the days before everybody had a computer. I would like to think that -- for example, the PKU program has been going on for 40 years. I would like to think that there's some long-term data that somebody has on the treatment of kids with PKU.

I know that relatively recently there was a recommendation about pregnant women. I assume that that resulted from women who are pregnant with PKU go back on the diet. I assume that that came from some long-term follow-up. Are you telling me that there is no long-term data on patients who have had PKU?

MS. TUERCK: Just from the collaborative studies that were done in the '80s. The maternal PKU study ended about four or five years ago, but prior to that the regular PKU study ended in the mid-'80s, I think. That's a good point, because right now there are some children with PKU who are responding to biopterin therapy rather than to dietary therapy that do not have known biopterin defects. That's huge. That's huge news. We can't even get the drug in this country.

But we are finally now beginning to pull together some clinical studies on this, and I just heard last week that fortunately we will be a part of those. But no, all that data is sitting in kids' charts around the country and nobody is pulling it out, because nobody has the time or the money to do it, or to do it in a consistent enough way that it is meaningful data for anybody. So the only way that we get long-term follow-up data on these diseases is that we have to wait for a particular doc or a particular group of folks to publish the data on their individual children. So you get four or five kids at a time, and that's the best we've been able to do. There's no national data on any of these diseases.

DR. HOWELL: Dr. Alexander, would you like to comment about the PKU studies since your institute funded these?

MS. TUERCK: We've got the expert here.

DR. ALEXANDER: NICHD funded the long-term follow-up of children born with PKU and their unaffected siblings, looking at the impact of early diagnosis and treatment intervention as a means of preventing mental retardation, and these were the studies that documented the efficacy of the screening, early diagnosis, early identification, and the effectiveness of treatment in preventing mental retardation. The kids' IQs were not significantly different if they'd gone on treatment early from those of their normal siblings.

These children were followed for a fairly long time, but the dollars just were not sufficient to continue those forever. Then we discovered the problem of maternal PKU, and Rod Howell chaired a consensus development conference that NICHD sponsored on maternal PKU, and we were able to get a significant number of women who had gone back on the diet at varying times, prepregnancy, early pregnancy, late pregnancy, not at all, to get a clear indication that the diet was extremely beneficial in reducing the likelihood that a child was going to be born with unalterable mental retardation if the mother had elevated phenylalanine levels during her pregnancy, and that the diet was effective in reducing that likelihood.

So those data, though, were gathered prospectively in a study that NICHD initiated of maternal PKU with a fairly long-term follow-up of the kids. Once again, those studies, they went on a long time. They went on for 10 or 15 years, and they provided the database that the panel used in reaching its conclusion about the effectiveness of a reinstitution of the diet as a means of preventing this as a problem. But they didn't go on forever, and those studies were stopped about five or six years ago.

DR. HOWELL: I think the issue is that there's huge investment in long-term follow-up, and I think that consistent with what Judy brings up is that these were ended before the clinical discoveries about biopterin. So there has not been a long-term follow-up. I think that that simply underlines the really critical need to follow these folks.
We could spend a month on this subject, Judy. Thank you for this. But we must go on to the data thing.

MS. TUERCK: Thank you.

DR. HOWELL: Let me also point out that if there are people in the audience who would like to make public comment tomorrow and who have not yet signed up downstairs, you must sign up if you would like to speak tomorrow. It's not clear. We'll have to ask tomorrow Dr. Puryear whether everybody will be able to sign up.
Dr. Hanson reminded me that he could send any member of the committee supplements to the published results from the consensus conference that was published in Pediatrics. If anybody would like that, Jim will provide those for you. He can send them to Michele.

But anyway, people who would like to talk tomorrow, sign up, and Michele will see whether or not the schedule will accommodate those people. We have a lot of people already signed up, but there might be a few vacancies.

We now need to move on, and we'll apologize to Dr. Ross that we're running a little late. But Dr. Ross, from the Public Health Informatics Institute, is going to discuss with us the overview of integrated information systems supporting child health in the context of a national health information network.
Dr. Ross?

DR. ROSS: Thank you.

That was really an illuminating discussion. I'm extraordinarily pleased to be able to be here to hear it.

I'm going to ask you to sort of come now out of the laboratory and look at the information world in a slightly broader context, but certainly a context that impacts the management of information about children to bring about better health and treatment outcomes.

But as I march forward through this, keep in mind something that Mark Twain said. He said the worst of all possible deaths is to be talked to death.
(Laughter.)

DR. ROSS: I think if he were alive today he would probably alter that and say that the worst of all possible deaths is to be talked to death about computer systems.
(Laughter.)

DR. ROSS: So we in this business have a challenge, to try to put what we think to be goodness in terms that the rest of you would care to hear. So I'll try to do a little bit of that at the risk of maybe being too general.

What I'd like to talk a little bit about is the national context. We hear an awful lot about electronic medical records, electronic health records, exchange of electronic information. Judy made a very, very important comment. She said why do we have to reenter the data? I think that's a really good question. It's a very important question. It's not easily answered. But there are things going on that may, if we stay the course correctly, help solve that problem.

She also said we have all this data sitting in charts. It's true. It's a huge national challenge to put that data together to make information that changes the way in which health practitioners function, the outcomes that actually come about with kids. So this is clearly a non-trivial and complex problem.

I'd like to talk a little bit about what's going on nationally and give you some data, a more simplistic way that we've looked at answering this question is there an information problem. I think we've already heard a lot of information that says there is. I'll talk a little bit about the activities in integrating health information systems that we have been doing very largely in collaboration with the Maternal and Child Health Bureau at HRSA for the last few years, and then end with talking about improving health outcomes through integrated information systems.

This recent Dilbert cartoon struck me as useful. "Our differentiated value-added strategy is transformational change." We hear a lot about that in the health care IT business these days. The boss says, "How was that? Does anyone feel different?" And then she says, "My urge to hurl has increased a little bit."

(Laughter.)

DR. ROSS: This cartoon makes a couple of important points. One is that we are hearing an awful lot of hype about what health care information technology is going to do for you. But I think there is also an awful lot of substance in that, and I would ask you to curb your urge to hurl.

(Laughter.)

DR. ROSS: I think we are at a time of opportunity, I believe. We know that complete information can impact health outcomes. The question is how do we make complete information? Public health has a great big problem. Public health agencies have disintegrated information. They have bits of information lying in different programs, for lots of good reasons, and this is not casting stones, it's just a fact. Within the average public health agency, we don't know what we know about a child. In other words, we can't create a unified record, what Michele Puryear has referred to as a child health profile.

In many places, almost all but in very many places, if a pediatrician were to call the health department and say what can you tell me about Johnny, it would become a pretty difficult exercise to answer the question. So I think at a starting point we ought to say is that acceptable? Our group is a non-profit organization, works throughout public health, and we are advocating to say the answer to that is no, that's not acceptable. We think that providers and parents have a right to complete information. So this is not a talk about all the technologies that potentially put that together, but I think we ought to keep focused on really what's the big goal.

Public health should be able to produce a consolidated record, and I want to talk a little bit more about the follow-up problem because I think it's a real one. Basically, the socko here for me is that you, the policymakers and the advocates, need to stay the course to continue to push that both public health and health care integrate their systems to the point where these data can move to providers in a timely way to put a complete and accurate data set about a child in front of a provider and to do it in a way that also assists parents to help do their job. It's an information problem that's endemic to the entire medical community and public health community.
So what's happening now? A lot of hype, a lot of visibility recently given to the appointment of Dr. David Brailer as a national coordinator for health care information technology. Since he has been appointed beginning last April/May, they've reframed the dialogue to refer to a national health information network, which is described as linking disparate health care information systems together to allow patients, physicians, hospitals, public health agencies and other authorized users to share clinical information according to stringent privacy and security protections.

Prior to that, we were talking about a national health information infrastructure, so this is an evolution, and that one was described more in terms of the technologies and the approaches, the need for standards, data standards and vocabulary standards to make this exchange of person-specific information possible. All of this came together last summer at the National Health Information Infrastructure conference, and a framework document, "A Framework for Strategic Action," was published by Dr. Brailer's office. It is beginning to lay out a national agenda, but an agenda that really is largely in the hands of the private health care world to deliver.
A corollary to the notion of this national health information network is regional health information organizations. Basically, we have on the health care scene today emerging organizations that seek to network together, and when I use the word "network", first I would use that as a social network, to bring together the health provider community within a region to agree that they want to do this. As they do that, put together the technologies that will make it possible. But this notion of a regional health information organization, you're going to hear more and more about that over the next few years. Several of these exist around the country and show great promise, but this is really an emerging aspect of our health care scene.

Finally on the public health side, for the last five years or more, CDC has been working, taking the major lead within public health agencies at the federal level, trying to develop a framework of standards that will allow and eventually bring about the rapid exchange of data that are relevant to public health. So it fits into this broader context of the national health information network. CDC refers to it as a cross-cutting and unifying framework. It is, in effect, an information architecture of standards -- data coding, vocabulary, message formats, et cetera.

So this is as much gig talk as I hope I do today.

Is there an information problem? What I'm basically telling you is that there is a lot of energy across the country right now to try to tackle this problem of assuring that complete data land in the hands of the providers when they need it, but it's not easily going to happen.

So let's look a little bit just at newborn dried blood spot screening and newborn hearing screening as examples of this information problem. We took 1999 data, and as you see, of the over 4 million screened, there were about 3,494 not normals. Of those, 154 were lost to follow-up, and 302 were classical PKU or clinically significant variants. When you take out the 154 lost to follow-up, you have 3,340 not normals

that were followed up, which is about one case per 11 followed up. If you apply that rate to the 154 that were not normal, does that mean we have 14 missed cases? I don't know. Probably not. But what it does say is we have an information problem. We don't know, and I think that is really the point here and we should be asking that question. Is it acceptable to just not know?

We looked at hypothyroidism. Again, over 52,000 screened not normal, 1,371 lost to follow-up. When you apply the same kind of logic, it leads to about one case per 32.8 followed up. Apply that to the number lost to follow-up and we end up with 42 missed cases. Is that appropriate arithmetic? I'm not sure. Is it telling us that, in fact, these kids were missed, or does this tell us we have an information problem? Again, we don't know.

We looked at days from birth to initiation of treatment. I think the big number here is the unknown. Is that acceptable? Is it acceptable for long delays or just not knowing?

There are a lot of barriers to gaining access to newborn screening results. Certainly, this list is a list that we've put together, one of my colleagues, Dr. Alan Hinman, who has worked very closely with us in all of this, put together. But there are issues of how we account for the way in which the health care system handles these data. We have found as we've gone around the country and worked with various programs that frequently we don't know some important information. We don't know if the laboratory received every single specimen.

I'll tell you, I think the newborn screening system of this country is one of the real high points of American medicine. I mean, I think as far as systems go, this is very good. This sets a quality standard. So when we get to some of these other child health systems, it's much looser.

So we have a lot to learn from the newborn screening programs, but transfers within a practice, the fact that in some hospitals, a very busy birthing hospital, may assign a single physician to many of these children. So getting the result to the right pediatrician in an absolutely confirmatory way, that we absolutely know it went correctly, is part of the information problem, and it's because we don't have the systems linked together in a real fully functional manner.

If you look at average time for the initial screen-positive result, again there's a fairly large percentage, over 5 percent, where we don't know. Average time for screen-negative result, nearly a quarter we don't know. Now, it's very interesting. When we talked to a lot of pediatricians, the general answer when asked do you know if the newborns that you're treating are screen-negative, their answer is usually if we don't get a result, we assume it's negative.

In a way, our health care system is working on the basis of a hope and a prayer. Is that acceptable? Do we want a system where we tolerate a situation where if the doctor isn't told, the doctor assumes it's probably negative? "They would have called me, they would have tracked me down if it was positive." Probably that's true, but this is an information problem that can be solved, I think, as part of this national transformation and automation of the health care system. So I'm not arguing today that we build a new categorical system just to handle newborn screening results, but rather let's make sure that it is thoroughly integrated into the overall automation of the health care system that's beginning to take place.

We looked at newborn hearing screening in Greensboro, North Carolina. About 3.5 percent of non-ICU newborns have abnormal screens. If you kind of go through the logic of it, you end up with a ratio of positives to confirm hearing loss is 17. The question is did any of the 18 that were not followed have hearing loss? So I pulled out some slides. This could have gone on about immunization registries as another way to look at some of the public health experience of making more complete information, but one of the things we've learned about the demonstrated usefulness of a immunization registry, the issue of registries has already come up today, and I think it's one we're going to hear a lot more about as we continue to automate aspects of health care.

But an immunization registry, where they are now fully functional and properly deployed, they have very useful results. Sending reminder and recall notices has an impact. Generating official immunization records. There are a number of aspects of having automated that piece of the information puzzle with children that's returned positive benefit.

So the basic premise here is that health services can be improved by assuring timely provision of accurate and comprehensive information. Our current situation is that information is often not timely, it is usually fragmented, and in the case of public health it is almost always fragmented. Of course, more money is needed to fix that problem. But do we need integrated child health information systems?

We know that many children do not receive all their preventive therapeutic services in a timely manner. Studies have shown that lower immunization coverage rates are also correlated with insufficient screening for lead and anemia. Multiple public health programs focus on the same target population without coordination of services and outreach. So again, I would ask you to look beyond the very specifics of newborn screening and think about the broader problem. It's a mistake for public health agencies to allow this disintegration of their information, the net result being to provide poor service, poor coordination outreach to patients that they are involved in the follow-up for.
Finally, I want to say that when we talk about integration, we're really talking about from the end user's point of view. It is time that we think about this in terms of the information put in front of the doctor. It needs to be complete. How you got there, there are lots of ways to skin this cat technologically, I think. But we need to have that as a goal, and keep the simple goal in front of ourselves.

Now, in our work with the Maternal and Child Health Bureau at HRSA, we have looked at systems potentially to be included in an integrated child health information system, and we break these into two tiers. The primary tier includes immunizations, newborn dried blood spot screening, early hearing detection and intervention, and vital registration. A second tier could include WIC, lead screening, EPSDT, and birth defect surveillance.

Now, why would we tier them that way? Well, the top tier really are held together by some common characteristics. First, they're recommended for all infants and children. They're carried out or begin in the newborn period. They're time sensitive. They're primarily delivered in the private sector but have a strong public sector component. They're mandated in almost all states. It would be my dream, since a few people today have offered their dreams, one of the dreams I would have is that as we reengineer the vital records system of this country, we do so with a change of attitude.

Right now, the vital records system that we have in this country is built basically out of 1940s thinking. It's built out of when we registered everything by hand on paper, and the notion of just recording in the Book of Life your name was the goal. We're way, way beyond that today. What we need to do is separate out this cluster of information that could empower other activities from the mere fact of creating an official record. There does have to be an official certificate, but that process can be somewhat separate from the process of feeding information to the newborn screening program.

Wouldn't it be nice if every public health laboratory had a daily electronic feed of every birth in their state so that they would know for certain that they have a blood spot on every child? Similarly, would it not be good that that electronic record of birth feed an immunization registry or seed an immunization registry so they would know that the first hep B shot was given to every child? This shouldn't have to be reentered and reentered and reentered. It's costly, it leads to error, and we're at the time now where I think the technology can actually fix it.

We're now also in the world of e-government, lots of e-government initiatives. The federal initiative for consolidated health informatics is going to have a big impact, I think, by creating standards that ultimately we will all live with. There have been important things done in the last year. Secretary Thompson announced the arrangement with CAP to make SNOMED licensed broadly, and the Medicare

Modernization Act will impact something with e-prescribing. So elements of the overall medical system are getting automated.

I've mentioned about Dr. Brailer, and I won't mention more about that other than to say that, of course, we're only at the beginning. There's been a very minor pitch for money. I think in the world of Washington, $15 million isn't a lot of money, but that did not happen. So I think we're looking to the private health care system to begin to build a business case that rationalizes why automation of health care is essential, to improve quality and safety, and ultimately also cost.

Now let me talk just briefly about the work we've done with the Maternal and Child Health Bureau. We've worked to facilitate the development of integrated child health information systems. Dr. Puryear earlier today mentioned the SPRANS grants. That has become a platform for us to work with them and work with a number of state public health agencies to define what an integrated child health information system is and to begin to understand how you go about building, designing, implementing such things.

That work led to this little book. It's actually in public health a best seller, and that's because it is written for the average maternal and child health program person to read and use, and it gives some case studies of public health agencies who have been successful at building these kinds of environments. It's available through our website, and if anybody ever wants a copy of it, you certainly can contact me and we'll give you a hard copy.

So what are some of the lessons? Very simple. First off, data are for sharing. That theme has emerged today and it needs to be really hammered on. Information increases in value the more it's used. We all know this fact of life, that information is not a rigid, hard asset like an old factory machine that you had to control its use because it would wear out. Sometimes we manage our information as though it was a machine that's going to wear out if we use it. In fact, this information needs to be used more. So sharing information to increase research knowledge, to empower practitioners, to inform parents is what we really ought to be about.
"Listen up" refers to the fact that to make this work, this is a sociological problem. It's a matter of communicating with many different constituents, and within public health agencies different parts of maternal and child health programs talking to one another, getting to know one another, working with one another, that personal handshake, that personal integration precedes system integration. Anyone who has ever tried to integrate systems and make it change the organization understands what failure is all about. It has to start with the people.

We've talked a little bit about change is hard. I also want to make another point, that where we've seen integrated child health systems function well, it's built on the fact that the programmatic people, not the IT people, are driving the train. You, the practitioners, the laboratorians, the epidemiologists, you really have to drive this, because the systems have to serve your purposes. I often say if you let the IT guys be in charge, you get what you deserve.

(Laughter.)

DR. ROSS: And finally, I think you have to stay the course. This is a long-term proposition. Integrating these systems will take at least a decade or two, and we have begun a useful process. We can't give up.

We are also in collaboration with the Maternal and Child Health Bureau working on principles and core functions of integrated systems. We've gotten help from a few people here in the room towards these work groups. We have developed a document that will be made public fairly soon. It's undergoing revision, so I really won't talk about it at this point to any great length. But we will move forward and actually try to define these systems in a way that the document becomes, in effect, a set of requirements that every public health agency and any private organization that wishes can have, make it a public good. I think if we do that we will eliminate some of the barriers to groups getting themselves more automated and implementing systems that actually integrate their activities. Right now it's largely a mystery.

We're also working on performance measures, and they have been developed with a national collaborative work group drawn heavily from the SPRANS grantees and that we field-tested this year. The issue of measuring performance came up earlier, and I want to say our philosophical bias, and that is that no information system, no clinical or public health information system should exist without performance metrics tied to it and actually measured. If we don't do that, how will we know if it's doing what it was supposed to do? If we don't ask the question to start with, how will we know what it was supposed to do to begin with?

We've had a lot of systems investments, under way, frankly, and when you really push hard and say what's this doing for you, nobody knows. We don't even know how we would measure what it was supposed to do for us. So we feel passionate about this one, that if we can't define how it's going to impact health status, why should we even start it?

Finally, we have been working through our All Kids Count program -- we have been a national program office for the Robert Wood Johnson Foundation for the last 13 years -- to build a community of practice composed largely of public health program staff but increasing -- I hope that over time it grows to bring in private provider community, to understand together what these systems need to do for us, what works, what doesn't work. This is an emerging field and this is a great opportunity for the idea of a community of practice, a way of managing emergent knowledge to help everybody grow together.

I want to thank HRSA for having been so supportive of this. We've migrated from our RWJ funding to having Maternal and Child Health Bureau help support this effort. Throughout, we will develop a business case for integrated information systems. Again, to kind of come back to that theme, we really need to rationalize the investment before we head forward with the investment.

So the history of this has been to develop a number of key lessons that are in your slide sets, and I don't think I'm going to hammer on them more than the very first one, and that is the involvement of stakeholders. These kinds of systems, as I said, it's a sociological problem. We have to involve everybody together in defining what they're to do and how they're to work and how they're to bring benefit.

Then the very last point here, number 10, use the information. My colleague, Alan Hinman, is fond of saying that the best is the enemy of the good. Until we use these data, they won't improve. But if we use them, they will improve, and the systems that support the data will be forced to improve.

This screen is not meant for you to read, thank God. What this is is a result of a survey that we did where we surveyed 19 health departments to look at the number of systems that they were working to integrate, and which systems, and basically this confirms something I was saying earlier, that newborn dried blood spot screening and hearing screening and vital registration and immunizations are on almost all of those states' lists. They're either actively working to integrate them or in the plans to do so, but they're also looking to integrate in children's special health care needs data, WIC programs, patient billing systems, birth defects registries, and early intervention programs.

So there is a lot of activity going on within public health, the goal of which generally is to bring about this more consolidated, complete record on children.
We have a supplement also available in electronic form via our website that a number of us participated in creating, a supplement to the Journal of Public Health Management and Practice. It was an issue focused on integrating child health information systems. If any of you want it, you can get it from us either in hard copy or electronic form.

To the issue of electronic health records, the American Academy of Family Physicians is really taking a very aggressive position in this. They've put forward the statement that every primary care provider will use information technology that includes electronic health records with the ability to access and communicate needed clinical information to achieve high-quality, safe and affordable health care. They did that in 2001. Their goal is to have at least half of their members using EHRs by 2006. The pediatric

community is I'd say lagging behind that, but I think the pediatric problem is, in fact, even harder to solve. We're not going to talk about that now, but if any of you would like to talk about that offline, I'd love to, because I think it's a complicated problem.

The American Academy of Pediatrics, they held a special meeting, I believe sponsored by HRSA, a few months ago, the goal of which was the adoption of electronic health records by the majority of AAP members in the next 5 to 10 years. So that's kind of the point of discussion. So we've got to get to where we actually have an electronic health record that is in fact a really workable, cost effective, supportable tool for pediatricians.

So what does all this mean for the future? I think there's a lot of activity going on. Most of it is focused on clinical systems. Some of it is focused on public health systems. It's geared towards more the programmatic functions and not so much the research functions. But these systems, I certainly think, to be cost justified have to support day to day work practice. I think it's to me an open question as to when they will actually be good enough to support the research community. Certainly probably in the not too distant future they'll support the first level of research questions, but I don't think we should get our hopes up. Let's first see if we can improve the quality of the way in which we give care through better information and then think more about research.

I think there's a great opportunity to assure that program systems support research endeavors, no doubt into the future, but researchers are going to have to be at the table. Right now I see these as separated a bit, and certainly on the public health side, epidemiologists need to assure that they can get good information from programmatic activity.

So how will health outcomes be improved or changed? I think the evidence indicates that improved, timely provision of accurate information in individual program areas has an impact. I think it's too early to demonstrate much of this impact, however. So there is something of a leap of faith on the public health side that integrating these systems can improve program performance. We're trying to develop the business case that would in fact argue that case in a more quantified and complete manner.
We need to do something about -- the current rate of loss to follow-up of abnormal hearing screens, for example, is unacceptably high. Children receive immunizations at birth, at 2, 4, 6, 12 and 18 months, providing multiple opportunities to act if information is available. So we have a system that doesn't coordinate well, and I think the information systems can support it. The early hearing detection intervention, EHDI, guidelines promote integration of systems. That's a recent grant guidance from CDC to the states, and I would applaud CDC for having included that notion of integrating systems as part of those grants.

I think with that I should end. I just want to say that we're all going to hear more and more about the automation of health care, and the interest in this room today ultimately I think can be served if you all remain a part of that dialogue. Thanks.

DR. HOWELL: Thank you very much, Dr. Ross.

Questions or comments? Steve?

DR. EDWARDS: One of the problems you alluded to with the follow-up of kids with positive screening tests -- I'm from North Carolina, and we have roughly 100,000 deliveries a year. We end up with about four children with PKU per year, and yet a couple have gotten lost out of that four. So what's going to happen when suddenly we're talking about numbers like 1 in 5,000 kids having some sort of abnormality based on the screening? How do you envision systems that are going to pick up those kids?

There are ways of doing it, being careful to take a lot of time. For example, in my office, if we didn't have the results back by two months, we called them on the telephone, which is very labor intensive. But the confidentiality issue comes into this, so there's no way that you can do it at this time by computer. So how

do you, with this integrated record, which I think is visionary and I think it's going to be real -- you pointed out some of the problems with the pediatric record that's unique to pediatrics. But how do you work through the problems of confidentiality with the need for information?

DR. ROSS: Well, you know, a decade ago I think the confidentiality issue was a major deal breaker. It was a social dialogue that had to start, and HIPAA and the legislation of HIPAA started that dialogue, in my opinion. I think that the technologies are going to provoke a discussion about privacy that will answer your question. Let me explain.

What technologies do is make explicit how confident we can be about something being secure, and it's going to take time before we as a society become comfortable with how much less than perfect security we can tolerate. In the old paper world, I used to be a hospital administrator, and I marveled at how our records were just sort of out there. Paper records, going to the hospital, they're at the nursing unit, they're just there, and people picked them up, and anybody could go through it.
But our protection was that, a, it was hard to find them. About half the time you couldn't know where was a patient's record to begin with at all. You just assumed it wasn't lost, that someday it would turn up, it was on somebody in the house staff's office, you know? Or you also lived with the sense that we don't know how insecure it is. So it's sort of another one of those 800-pound gorillas that we just didn't want to look at.

Automation makes it very, very obvious. So now we have to answer it in the specific, but the technologies are I think pretty much there today, and if they're not totally there, they're going to be, that make the ability to make people's individual personal medical information very secure because we will be able to know who looked at it, when they looked at it, and potentially even what they did with that information. We still, though, have to make a leap of faith that we're willing to put rules in place that guide our behavior when we exchange and share that information.

I think as the society grapples with that, how comfortable we will be, ultimately we're going to reach the point when we say there's more to be gained by sharing the information than lost in keeping it isolated. As we do that, they're going to take case studies, case examples where improved outcomes come about because you've now coordinated different aspects of the health care system, or that you've made it easier for public health, for example, to do things for you that as it currently exists they're putting the burden on your shoulders to say we haven't heard about this kid, let me go track them down.

I think the information systems can solve many of those problems, but the social dialogue isn't yet complete, in my view, to make it possible. It's complete within corporate health care entities, single entities. The regional health information exchange organization that I referred to, that's going to force the dialogue about what are the acceptable rules for one corporate health care entity to shift information to another or share it with another, and that's evolving. I don't know.

DR. HOWELL: Bill?

DR. BECKER: David, I think that security is the key issue, and I'm glad you touched on the regional sharing of data, because from the states' perspective, HIPAA does not prohibit the reporting of public health mandated data. So in other words, it's not a confidentiality issue for the state to request data about public health mandated testing or reportable infectious diseases or any of the other programs that the state operates. But that data needs to be secure -- that's a huge issue -- and the sharing of that data on a regional basis is an issue that absolutely has to be tackled.

I have two questions for you, which seems to be my popular thing here today, two. The first is, when last I was briefed on this particular topic, I understood that there were a couple of states, and I'm thinking Missouri and Georgia, and I know there were about four or five others, that were trying to develop some integrated data systems in their health agencies with all the items -- immunizations, vital stats, WIC, et cetera, newborn screening -- and I was wondering if there was one out there, without getting into the gory

details, is there some evolution or some state of evolution of an integrated system out there that you think is showing some promise?
Then my second question is have you looked at Judy's database, and what did you think of that?

DR. ROSS: I'll answer the second question first. No, I have not looked at Judy's database, and I'd love to look at it and understand that. I was fascinated, but it's basically new to me.

In answer to your first question, there are a number of states that have tackled this notion of integrating child health information systems. They've all tackled it from different perspectives in different ways. The work we're doing with the Maternal and Child Health Bureau I hope ultimately gives a detailed and very clear road map to the states as to what they're embarking upon and what the requirements are, the technical requirements are to build such a system.
Right now we are having everybody try to reinvent that wheel themselves, and I don't think they need to, sort of like what we did with the public health laboratories. Why not let's work together, develop that set of requirements together as a base document, and then let other people embellish upon it rather than let everybody try to start from scratch and develop it themselves?

As far as states that have made a lot of progress, there are a few, but the state that really comes to mind is Rhode Island. They've had a system called KIDSNET for a decade or more. They're creating new technology platforms for it that are going to be much more functional, but that state is also a leader in that they are one of the five states that received an AHRQ contract this past year to enter into a collaboration with a regional health information organization. In Rhode Island it's called the Rhode Island Quality Institute, and the public health agency there is a key partner in that non-profit entity. So lots of new ground is being broken here.
Dr. Pat Nolan, I think to her great credit, realized that the vision for that Rhode Island Quality Institute is to improve the health of all the citizens of Rhode Island, and therefore the health department needs to be sitting at that table. So she is actually vice chair of the non-profit entity. But the Rhode Island Department of Public Health is using KIDSNET as its data system that will be added into the integrated environment. So they will be allowing private physicians in the various hospitals and private practices of that state to gain access to the KIDSNET data.

So we will see at least one situation where the provider is able to get, with some initial problem but ultimately relative ease, a complete record on the child. So they'll see up-to-date information. That's a social experiment that's just beginning, and I think we're just going to have to monitor it. But it's one of a handful.

DR. HOWELL: Thank you, Dr. Ross.

I think that we're right on time. So let's take a 15-minute break, and we'll return at 3:15.
(Recess.)

DR. HOWELL: We're going to proceed with the afternoon activities, and there's been considerable interest in the update on the American College of Medical Genetics' report to HRSA, and Dr. van Dyck will take us through that situation.

DR. VAN DYCK: Good afternoon. The committee reviewed a very early draft of the report and recommended at the last meeting that the report go forward to the Secretary with a letter from the Chair, Dr. Howell, transmitting the report to the Secretary. The committee also said that report should be sent after it's fully edited and complete, with accompanying public comments from the meeting.

The report has been completed in the last week. It now has the appendices added. There's about a 200-page appendix which contains -- and I just want to help people understand what wasn't there when they reviewed the draft. It contains a two- to three-page description of every one of the disorders that are discussed in the report, so 80 or 90 different conditions, and on those pages -- I have one for MCAD in

front of me -- it talks about the type of disorder and the ethnicity specifications of those children who may be at risk for the disorder, the way it's screened for, how many states in the United States screen for it at the time the report was prepared, and the percent of births as of August 2004 that are screened for that condition in the United States.

It talks about the incidence, and then there's a literature cite for where that comes from, a phenotype at birth and another literature resource, and the burden if untreated, also again with another literature resource. Then it goes into the test and talks about the screening test, the overall cost of the screen, what are the multiple analytes that are possible when you screen for that particular test, the secondary targets, there's a description of each of those and then there are literature cites for each.
Then the same for the treatment, availability and cost, efficacy of treatment, benefits of early intervention, benefits of early identification, prevention of mortality, diagnosis, acute management and simplicity of therapy, with a discussion of each, a short discussion, a sentence or two, and then the literature cites for those references.
Then on the second page are the literature cites, a comment in general about the disease, and included with all this is its score in the report and its recommendation to be contained in a core panel or not. As far as I know, this is the first time that this kind of full information has been available ever for even a handful of these conditions, but now it's in one place for all of them. This took a little longer than we planned to get this all together, but we do have a completed report that's been completed in the last week.

Now, the process for the report. We want to get the widest possible public input we can for the report now that it's complete, and so we are working on getting departmental permission to release the report for public comment. We'll probably allow about 60 days for the public comment period and certainly for the committee to comment again as well. We will then, in the Department, take those comments, all the inputs, the input from the report, inputs from other meetings such as these and from the committee, and try to put that together into some kind of a recommendation from the Department.

Now that the report is done, we also feel comfortable giving it to the committee for its transmission, not giving the report to the committee but appending the report to the letter from you that goes to the Secretary. So I think we're ready to solve both those loops. If people have questions about that process, I'd be happy to try to answer them.

DR. HOWELL: Any questions? Bill?

DR. BECKER: Peter, do you have any idea or guesstimate about when it would be posted on the web or made publicly available? I mean, if you don't, that's fine.

DR. VAN DYCK: Well, I don't want to speculate, but we're hoping very soon. It's to our advantage to get it out as soon as possible.

DR. BECKER: Will there be announcements?

DR. VAN DYCK: Yes, I think we will announce it and we will certainly send it out for public comment to those affected agencies and folks, and of course to the committee.

DR. BECKER: That would be great. Thanks.

DR. HOWELL: And as I understand the process, I've actually drafted a letter that will accompany this report, and it's my understanding that that will go forth now, this report, with my letter as soon as it's available. Is that correct, Peter?

DR. VAN DYCK: Yes.

DR. HOWELL: And at the same time the thing will be posted for public comment.

DR. HOWSE: Peter, I'd just like to ask a couple of questions but begin by making the statement that I made this morning, which is from the standpoint of the March of Dimes. We would like this report to be in the public domain as quickly as possible so that the public and all the interested parties that have been waiting so long for this report to come out -- professional groups, parent groups, state leadership -- can have an opportunity to comment, to read and to comment on the report. So that's the first statement, that this report get into the public domain as quickly as possible for public review and comment.

DR. VAN DYCK: We agree.

DR. HOWSE: Good, that's good.
As a separate but related matter, the moving ahead of this report within the Department, I just want to say for the record that I think, having read the minutes of the last meeting -- I didn't attend the last meeting, but having read the minutes and spoken with some of my fellow committee members, I think a substantial number of us on the committee were of the impression that the recommendation that was made by the committee in September, which was that the report be forwarded to the Secretary's office and released into the public domain, that we're disappointed that that didn't take place soon after the September meeting. I mean, it's very clear that that's what the committee voted to recommend at the September meeting. That's four months ago. That's a long time.
So I think that the concern will remain until the report is released into the public domain for comment and until we receive copies of the transmittal letter to the Secretary under Dr. Howell's signature and a copy of the report for this committee to review and begin its own public commentary process. We haven't even had a chance to comment on this report as the Secretary's committee.
So, three things, releasing the report in the public domain, we'd like to get a copy of the transmittal letter, and the final report as you described it for us as quickly as possible, and then to institute some kind of process for this committee to review the final report and the public comments that come in so that we can responsibly make whatever collective statement we want to make to the Secretary's office in connection with the process. So I'd just be interested in your response to those items.

DR. VAN DYCK: Well, I think that's what I just outlined. The only thing I can't tell you is the day that the report will be available, and we hope it's very soon. As soon as we have Department clearance, then it will come out for public comment.

DR. HOWELL: Bill, and then Steve.

DR. BECKER: A couple of issues were raised at the last committee meeting and brought out in the minutes that talked about the need for perhaps a slight revision to bring more of the peer-reviewed data into the commentary, and I'm wondering if some of the delay was due to some revisions to reflect -- well, actually, I'll just ask the question. Were some of those -- and I think some of the comments that Coleen made about the weight of the evidence that supported some of the recommendations coming within the report. Were some of those comments or concerns that were expressed at the last committee meeting, were those addressed in the final document?

DR. VAN DYCK: I think the College felt that they gained from the discussion in the committee, and that became part of the editing process, yes.

DR. HOWELL: Having seen the report through several things, the report has been substantially amplified with a lot of the data that was not in the original report, and there were comments from this committee that clearly have been incorporated into the text and so forth at the current time.

DR. VAN DYCK: I might add, I'm not sure the report is delayed, per se. I mean, you used the word "delayed." All of these fact sheets and all the rest were all a part of the original task order. What you reviewed was a report before those were done, a draft before those were done. So those were always in the process of being developed, and they've now been completed.

DR. HOWELL: Dr. Alexander?

DR. ALEXANDER: Relevant to that, my recollection from the last meeting was that while we did vote to forward the report to the Secretary quickly, it also was with the understanding that it would be the completed and final and edited report and not the draft version that was still pretty rough that we had some comments on at that meeting. So the motion carried with it the intent and understanding that what would be forwarded by Dr. Howell in his role as chair of this committee to the Secretary would be the edited final report, not the version that was still very preliminary and very rough that we saw at the meeting.

DR. HOWELL: That was my understanding, and I think that might not be reflected in the minutes and so forth. But at least there was a feeling, certainly members of the committee had some substantial concerns about certain parts of the thing, and it was felt that it would be benefitted by making those additions and so forth.
Steve?

DR. EDWARDS: I assume that with your optimism, Peter, that you don't feel that this is going to have to await the confirmation of a new Secretary, approval or disapproval.

DR. VAN DYCK: No, I'm not prepared to say that. I think confirmation has been scheduled for next week.

DR. HOWELL: Any more comments about the report? As I understand it, Peter, you can say that that's not what you said, but it's my understanding that you're optimistic at this point since you have the completed report at this point in time, that getting the ball moving and getting it in the public arena, which I think Jennifer has correctly pointed out is a critical issue, and getting it to the Secretary will really be prompt and we're not talking about months, we're talking about days and weeks. Is that correct?

DR. VAN DYCK: I surely hope so. It's to all of our advantage to have it completed as quickly as possible.

DR. HOWELL: Well, put it on your calendar and keep your watch going here, because I think it really is great anxiety to --

DR. HOWSE: What would be the method for getting the report into the public domain? How will that be accomplished?

DR. VAN DYCK: It will be on a website.

DR. HOWSE: Good.

DR. VAN DYCK: We'll send it to organizations who have asked for it or have an interest. It will come to the members of the committee. It will be posted on the genetic committee's website, as well as HRSA's and the Bureau's website. Every way we can think of.

DR. HOWSE: Great. And you think you'd like to see a 60-day public comment period, or do you think it will take more than that?

DR. VAN DYCK: No, I would think probably 60 days, but I don't want to be held to that because others may have a suggestion it be slightly longer or slightly less. But I would think a 60-day range would be appropriate.

DR. HOWSE: Fair enough.

DR. HOWELL: And since the document will be a public document, everybody will be encouraged to be sure to share it with anybody that has an interest I would think. There's been tremendous interest in the report, as you very well know. So I think people will be excited to see the thing surface at this point.

DR. HOWSE: It's certainly the best kept secret non-secret.

(Laughter.)

DR. HOWELL: I'm unaware of anyone in the country that has not had the draft.
(Laughter.)

DR. HOWELL: If you know of anybody, please let me know.
(Laughter.)

DR. HOWSE: I'm just trying to put some decency to the process now.

DR. HOWELL: It's a carefully kept Washington secret like usual, and as usual everybody knows. Maybe the best idea would be to say that this is a secret document, because in the previous case that ensured that everybody had it immediately.
Are there further discussions? There's been a tremendous amount of anxiety about the apparent slowness of this getting through, and I think that the amount of effort that the College folks spent in getting these final things together is enormous, and I think you'll appreciate that when you see it, and that took longer than one might have anticipated. I think then that HRSA was interested in editing it and spiffing it up, and I think all those things contributed. But hopefully it will be a better document once it gets out there, and certainly it has a lot more data in it than the bare bones early draft that everyone, except my cat, had a copy of.
Are there any other comments about this erstwhile report?
(No response.)

DR. HOWELL: Any other questions of Peter? I think he got off awfully easy. There must be something else.

DR. VAN DYCK: I just want to mention briefly counting conditions, only because it came up this morning, just to briefly say we're talking about a way again to bring some relevancy and uniformity to the way we count conditions so that we can score more carefully states for the public, and to try to eliminate the gaming and the competition. We are using the report as the basis for that, so this is not a separate exercise, Jennifer, as you mentioned earlier. It's a continuing exercise, as I see it, and will be brought to this committee as soon as we have something that can be discussed.

DR. HOWELL: Any more questions or comments?

PARTICIPANT: My question is has the vote been changed (inaudible)?

DR. HOWELL: Can you come to the microphone, please, so we can hear you clearer?

PARTICIPANT: Yes. My question is has the vote been changed from the 30 days that was the public comment period to 60 days during this period? If I remember correctly, it had been voted that there would be a 30-day comment period.

DR. HOWELL: The original vote had been a 30-day comment, and the group has not revoted on that. I think the issue that's percolated through is some of the federal regulations that maybe we're not fully aware of, frankly, and apparently 30 days is extraordinarily short. But we might discuss that again if you would like to.
Peter, could you comment on that?

DR. VAN DYCK: Yes, I can comment on that. The recommendation from the committee was that the committee allow 30 days of public input into their process before the report be forwarded to the Secretary. That's a separate process from the public comment once the Department releases the report. That's a separate process. The vote last time was for the committee to accept public comment from its constituents.

DR. BOYLE: It might be helpful just to clarify what we voted on last time as a committee, and just see how

this coincides with that. I'm fine with the process you described, but it is different than what's reflected in the minutes.

DR. HOWELL: Would someone like to make a recommendation on the subject? Coleen?

DR. BOYLE: Sure. I recommend that we clarify what we voted on last time.
(Laughter.)

PARTICIPANT: What page is it on?

DR. LLOYD-PURYEAR: Page 32.

DR. VAN DYCK: So the minutes really aren't right, then.

DR. BOYLE: I could just read the sentence if you want, okay?

DR. HOWELL: Okay. Why don't you read it?

DR. BOYLE: Sure. It says, "It was decided, given the strong interest in forwarding the report to the Secretary as soon as possible, the committee will accept and recommend the report and forward it to the Secretary immediately. There will then be a 30-day period in which electronic written forms from the public will be collected and the committee will form its comments and recommendations and forward them to the Secretary with appended public comments."
So it sounds like we were going to forward the report, put it out for public comment, and then take those comments back, including our own comments, and forward additional recommendations. Not exactly how I remembered it.

DR. VAN DYCK: Actually, what's going with the report are the public comments from the meeting last time, because that's what we understood the discussion of the committee was, with any additional comments that came in in the next 30 days. So that's what's been prepared to be appended to the report when it goes to the Secretary. Now, if somebody feels the minutes say something different than that, then I guess we need to know that.

DR. HOWELL: Any wisdom?

DR. BOYLE: I guess I just was a little unclear what our next steps were. I'm fine with what Peter described. After the 30- to 60-day comment period, after the report has gone to the Secretary with the appended public comments from the previous meeting and anything that's been collected in the interim, then what's the next step? It looks like from what's written here, we said that we would take the comments from the comment period and form a recommendation based on the revised report and those comments.

DR. VAN DYCK: I see that being during the official public comment period that begins when the report is released, that the committee will then take the comments that are received during that 60-day or 45-day or 90-day period and review them and make whatever recommendations the committee feel are appropriate.

DR. BOYLE: We're saying the same thing, then.

DR. DOUGHERTY: It looks like the committee is sending the report to the Secretary twice. I mean, that's what it says. "The committee will accept and recommend the report and forward it to the Secretary immediately." Then there will be a 30-day period with public comments, and the committee will form its comments and recommendations and forward them to the Secretary with the appended public comments. Can I make a recommendation that whoever takes the notes and we have an actual formal statement of what we're voting on, that those be in the minutes as they were stated, as we voted on them, rather than a summary?

DR. LLOYD-PURYEAR: This is directly from the transcripts. It was confusing.

DR. COGGINS: But didn't we also say that we should not send the report in draft form, that the report would be finished. If you're saying that we take the public comments from the committee meeting last time, plus the intervening period of time, isn't that 30-day period finished? If the report is being finalized, there's some public comments appended to that, it then goes to the Secretary, then it's available for a period of time for public comment, whether that's 30 days, 45 or 60 days, as you said, yet to be decided. But I don't think we took an action to send the report to the Secretary as a draft form. It was going to be finalized.

DR. HOWELL: That certainly was my impression, and I guess the problem is that this is from the transcript, I might point out, which means that our verbiage was not as clear as it might have been, clearly. But the bottom line is that it would not be prudent to send the Secretary a draft report that had not had these things incorporated in it. I think we generally agree about that. I guess that the key thing we need to do now is to clarify exactly where we are so that there's no confusion about where we're going from this point forward, frankly. Would anyone disagree with that?

I mean, it seems to me that how the minutes are worded and so forth, and what we expected and what we thought is kind of past history. But the bottom line is that we have a clear understanding of where we've been. We reviewed a draft report, and there was a good bit of discussion from the group about that. Some written material came from the committee. Those were incorporated into an updated version that has additional information, and that's been percolating along over the past few months. I'm told that that's now complete from Peter, and that as soon as he gets clearance upstream, that that will go to the Secretary and be put in the public arena for public comment from everybody.

Then after a period of public comment, those comments will come back. Is that not correct, Peter? And then we can review those comments and comment further about that. Is that not where we are?

DR. VAN DYCK: Yes, I agree with what you just said.

DR. HOWELL: Was that clear to everybody?

DR. EDWARDS: Well, I wasn't here, but I think that we need to take it from this point. It seems to me that that's a rational thing to do, to take it from this point and move
forward and maybe not -- I don't think we're solving anything by discussing some nuances in the discussion last time.

DR. HOWELL: I agree. I think the thing is, I think there were some differing opinions of what we were expecting.
But Peter, you had some comment?

DR. COGGINS: Yes. I was just going to say I think that is the rational way forward. I think the only thing that's left open is that period of public comment. How long is that? Is it 30 days, 60 days, or somewhere in between?

DR. HOWELL: There are federal precedents for these sorts of things that I certainly am not expert about. Peter, you were thinking that you were recommending 60 days, but you also felt that the agency or HHS may have differing opinions about a longer time. Is that correct?

DR. VAN DYCK: I'm suggesting it's most likely it will be 60 days, but I'm not promising that.

DR. HOWSE: Just to follow along what Steve said, perhaps what we could do is vote on a new motion about the disposition of the committee and the report, because I don't think we're going to settle the ambiguity of the minutes, and I wasn't even at the meeting so I don't even have any business attempting to be part of that process.
So as I understand what we've agreed to, there's a three-part recommendation that we would make, the

committee would make. Number one, the report will be immediately placed in the public domain for review and comment. Number two, the committee will review the final report and add its comments and recommendation within -- I don't know how many days you all believe it will take us to do that, but let's say 30 days so we can set the example for speed. And then three, HRSA will then transmit the report plus public comments to the Secretary's office for review and disposition.

You can't submit the report necessarily to the Secretary. I mean, the Secretary is going to say interesting report; what did the public think? I mean, I would hope the Secretary would say that. So if we do a three-part process that gets it into the public domain to begin with, the committee receives it through the public domain like everybody else, because we obviously don't have a copy today and we're going to have to wait until it goes into the public domain to officially have a copy of the report, we review it as a committee, make whatever comments and recommendations we want to make as a committee, and then that plus whatever the public has to say gets transmitted by HRSA to the Secretary for disposition by the Secretary's office, which includes a whole bunch of other layers of review. I mean, the Secretary is going to ask a bunch of agencies to take a look at it, I would think.

But meanwhile back at the ranch, we've got this report in public domain. This committee has looked at the final version of the report and made its comments and recommendations, and then the report is released by HRSA to the Secretary's office with a letter of transmittal which says here's what the public thinks, here's what your committee thinks, Mr. Secretary, that you appointed, and that process takes however long -- I don't know how long that process takes, but however long it takes, it takes.

DR. EDWARDS: I thought I heard one more step with that, and that is that the committee would also review the public comment. So if we could add that as a fourth, then I will second that.

DR. TELFAIR: This is a point of order here. If you're looking at the minutes -- this is sort of Roberts Rules, so forgive me for this. But if you're looking at the minutes and you're about to make an amendment to the minutes, you should be voting an amendment to the minutes, not as a new vote but an amendment to the minutes before you get going. You have to agree to that first before you can actually make the change. I'm just bringing that up. I don't know if that's what you're doing or not.

DR. HOWELL: (Inaudible) and that we're going to say that's there and that we need to move ahead and so forth. I think the one thing that I'm a little concerned and confused about is whether or not the report could go to the Secretary before the public comment or not, Peter. I don't know the rules there, how that happens.

DR. VAN DYCK: Well, the committee can do what they would like. It's up to the committee to choose what they would like to do.

DR. HOWELL: Dr. Alexander?

DR. ALEXANDER: I think that the process that was outlined by Peter and Rod is consistent with the minutes and with due process for both the government part and the committee's part. If, in fact, we go ahead and now that we have the final document and it's ready for release, Rod in his capacity as committee chair can fulfill the directive from the motion at the last meeting and forward to the Secretary with a cover letter from him in his capacity as chair this final version of the report from the College, prepared under contract to HRSA, with the comments that have been received from the committee at the last meeting and the other comments that were received within the 30-day period specified in the minutes accompanying them, just commending this report to the Secretary and urging him to act on it as quickly as possible. That was the gist of the minutes and the motion, and that would carry out the directive from the committee at the last meeting.

Then we begin the new process, and that's what Peter outlined. The report will go, in accordance with regular channels that these reports go, to the Department. We would propose that the report be released to the public for public comment for 60 or whatever final days the determination is for that comment period. Those comments would come back to the Department. This committee would have an opportunity

to be one of the commentators on the final report, which we have not seen yet, and be among the comments received by the Department on this report to be taken into account and forwarded to the Secretary.

If the committee wishes to see the additional comments and have a role in making further recommendations, we could request that. I don't know what the process is for making that kind of a determination, but that would have to be a request from the committee because that's not the usual process, and then the Department would have to decide whether they want to make all those other comments available to the committee to take into account and get their committee's input on as well.

But I think we are perfectly okay in proceeding in accordance with that outline, and we will fulfill the directive from the motion that was made at the last meeting. We don't have to change the minutes. We just have to agree that we're going to proceed as outlined in dealing with this final version of the report and the public comment process, of which we will be a part in terms of making recommendations to the Department about future actions. So I think it's all consistent with process.

DR. HOWELL: And that differs slightly from your thing in the fact that the report would go currently to the Secretary and into the public arena, and that we would comment to the Secretary after we've reviewed it with the rest of the public and so forth. I think that's the difference, as opposed to --

DR. HOWSE: You know, either way is, I think, fine. What we have today that we didn't have at the September meeting is a final report. So that's a huge difference for us as a committee. So I think what Dr. Alexander -- if I understand what you're saying, you're saying fulfill what appears to be the intent of the September minutes. Take the final report, which Dr. van Dyck says is final, Dr. Howell says is final, so there's a final report, take the final report and transmit it to the Secretary. Step one.
PARTICIPANT: (Inaudible.)

DR. HOWSE: From Dr. Howell.

DR. HOWELL: Right.

DR. HOWSE: Step two, release that final report into the public domain so that there can be an appropriate comment period. Step three, this committee reviews the report, the final report in the public domain and makes its commentary, and that's also sent to the Secretary under Dr. Howell's signature.

DR. HOWELL: Right.

DR. HOWSE: Step four --

DR. ALEXANDER: Here's where we differ. If you wait to do that, you're not going to send this report to the Secretary right away, as you had asked to be done. If you participate in the public comment period along with the rest of the public, you will have a second opportunity to comment on the final report and get those comments from the committee to the Secretary for consideration. If you wait until that process has gone on, Rod's letter is never going to get there.

DR. HOWSE: Well, how can this committee in good faith ask Dr. Howell to construct a letter, send the report to the Secretary for a report that we've not seen the final version of?

DR. ALEXANDER: That's what was voted on at the last meeting.

DR. HOWSE: We voted at the last meeting, I think, to send what we saw, which now is being called a draft, I think.

DR. HOWELL: Wisdom? Derek, we need an attorney over here.

MR. ROBERTSON: To be honest with you, I think there was some discomfort at the last meeting about

this whole issue, and I think it was really going to be a report that -- I mean, I think the minutes are actually kind of correct. It said there was some discussion about sending it forward. However, because we wanted to get it there and there was some debate about that -- but I think the earlier discussion is also relevant now. It's somewhat of a moot point. The report didn't go to the Secretary. So we just have to move forward, so how best do we do that? I think Jennifer's point has to be well taken.

As much as I want to see the report get there, you can't really in good faith send a report and say here's this report and the committee hasn't seen it. I know some folks had some discussion about the amount of data that went into some of the conclusions. We haven't seen if that's been changed or if that's been addressed, so some committee members may have an issue with that, that they want to comment on. I think you really don't have much choice.

You have two choices. Either you're going to send the report saying, well, the chairman has seen it and we're going to go based on what the chairman has seen. We can do that and say we have confidence in the chairman's judgment and go ahead and do it, or we see the report before we send it out. I would opt to say I don't think the main report has changed significantly, and I would go with the chairman's judgment.

DR. HOWSE: I'd sure support that. I mean, I think that allows all of our fervent wishes to be fulfilled. The report makes its way timely based on our confidence in our chairman, it makes its way timely under Dr. Howell's signature to the Secretary's office. Secondly, our fervent wish is that this be released into the public domain so that there can be public comment, including ours. So our second fervent wish is met by this approach. Then the third part, which is actually two parts. We make our comments and we also review what the rest of the public comment is, and we see where we go with that discussion. That seems to fulfill everyone's wishes in the matter and also demonstrates our never-ending confidence in our chairman.

DR. RINALDO: But the one question I have is I don't see the purpose of the first step. So we review it, and what?

DR. HOWSE: Well, the first step is just to get it in motion in review by the Secretary's office, just to get it in motion, to say yes, we think it's appropriate for the chairman of our committee to draft a cover letter which transmits this report on our behalf to the Secretary's office, and that simply gets the clock ticking on the review process within HHS and the Secretary's office, which can take a very long time.

DR. RINALDO: My question was strictly about the third point. I'd like to think that any action should have a purpose. So what are we supposed to do? Just review whatever other comments? It could be a thick pile. And what would be the outcome of that review? That's what I'm missing.

DR. HOWELL: Why don't you speak first, Joe, and then we'll hear Steve.

DR. TELFAIR: That actually was also a question, but mine was slightly different than yours. It was who is going to be responsible for the integration of the comments from the public, as well as the committee, and then the next step? It seems to me it's the step of having someone look at that integration before it's forwarded that is important.

DR. HOWELL: My impression would be that the public comment, we will see that, but we will not do anything accept transmit those to the Secretary. We will not integrate those. Those are not for us. Then the committee's comments, once they go forth, then obviously the committee would need to make comments, and we'd need to see a draft of the comments that went forth from the committee. But I would say that the committee will make comments on the final draft, and then those will go to the Secretary's office, as I understand what's being discussed around the table.
Steve has something.

DR. EDWARDS: My recommendation was that after all the comments are in, that at least this committee look at it again. We could amend our report or we could do whatever we wanted, but the recommendation

is that we present it to the Secretary now, not as a draft but as a report. But I still think that it would be inappropriate for us as a committee not to look at the public comment and to see if we would modify our recommendations.

DR. HOWELL: I think that we should see the report again with all the public comments and all the committee comments. I think I agree with you.

DR. TELFAIR: Then it moves forward again, and I repeat myself. If you do that, then somehow or another there needs to be some level of synthesis or some level of bringing together all the committee's comments into some kind of cohesive document, or else -- I don't know, I've not been on the committee. This is my first time, so I don't know what format it's in, but there should be some kind of synthesized format such that it is representative of this committee.

DR. HOWELL: What will the Secretary do as far as including these comments and all, Peter? The Secretary's office will obviously have the final say about all of this.

DR. VAN DYCK: Well, when public comments come back to the Department, they come to the Department and they are read and considered and appended to whatever report goes forward to the Secretary at that point, with recommendations from the agencies within the Department.

DR. HOWELL: And the document will go, as I understand it, the Secretary will send it to all the constituent agencies for comment. I believe it will go to the CDC and --

DR. VAN DYCK: I would think he would send the report and the appended public comments.

DR. HOWELL: To CDC, NIH, AHRQ, everybody.

DR. VAN DYCK: Right.

DR. HOWELL: For specific comments and action.

DR. VAN DYCK: Right.

DR. EDWARDS: I'm still stuck on one thing. I think the report that we send now is our report that we're sending to the Secretary. But at the end of this process, I'm suggesting that our committee look at the comments that come from other people and just make a decision about whether we want to make any further recommendations or not. Our recommendation, as I see it, would be what we send right now to the Secretary.

DR. HOWSE: May I try?

DR. HOWELL: Please.
Jennifer is going to synthesize all this extraordinary wisdom that has been coming up.

DR. HOWSE: So perhaps what we're emerging towards is a three-part process. I have your piece of it, I promise, Steve.

DR. EDWARDS: I'm not wedded to it.

DR. HOWSE: The three-part process. Number one, the report is immediately transmitted to the Secretary under our chairman's signature. It's the final report and it's transmitted to the Secretary under our chairman's signature.
Two, the report is released into the public domain for review and comment for the appropriate comment period, whatever that might be.
Three, this committee transmits its comments to the Secretary, which include the results of our own discussion, as well as a review of all of the public comment that is made.

DR. HOWELL: And that was in the form of a motion.

DR. HOWSE: Well, I was hoping for some expression on faces before I put it forward as a motion. (Laughter.)

DR. EDWARDS: My puzzlement with that is that I think we can make our commentary, but I'm not sure that we can, as part of the same process, make our commentary and then make our comments based on others' commentary, because our commentary, as I would see it, would come in the same time frame as everybody else's does. But then as a later phase, we would look at what other people have said. I'm not wedded to any of this. I think the process you outlined is fine. If that last part is sticky and nobody else thinks that we should look at it again after there's been public commentary, that's okay with me. But I personally think that we should.

DR. RINALDO: My confusion in this process is that we send what mix? The public with the committee? I'm not clear how that is supposed to happen, and it should happen. So the committee had a review, there was a vote, there were some concerns, and then there was a non-unanimous vote which is basically on record, but now are we saying -- to me it sounds like we send in our report and we kind of leave room for later saying we change our mind or something like that. I just don't understand it.

DR. EDWARDS: What I hear stated is that we have not seen this final draft. I haven't, and I don't think anybody has. So we have the same option as individuals, not as a committee, but as individuals to comment on it just as the public has an opportunity to comment on it.

DR. RINALDO: So those will be comments coming from individuals, not identified as members of the committee.

DR. EDWARDS: I don't think that's a critical factor, but I think that since we haven't seen the final draft, that the members of this committee should have an opportunity to comment on it just the same as any other public person.

DR. VAN DYCK: I think that's an important process for the committee. I mean, the committee should review the final report, and the Department would anxiously await those comments from the expertise around this table. I think that's an important public comment review.

DR. HOWELL: Peter has been quite anxious over here.

DR. COGGINS: I didn't want to complicate it, but just to Steven's question, if the report is so close to being finalized, why couldn't it get circulated right now to the committee members to have a quick review and pass on any comments? Then when it goes to the Secretary, comments from the committee can be included. Then you go to the public period and just append their comments following that. I don't know how close this is to being released, but the impression is it's very close, and I would imagine any further changes are going to be relatively minor.

DR. HOWELL: Any comments on that suggestion? The only point I have is the report is vast, and a quick look at it is not going to be possible, I don't think. I mean, you either measure it or something, but as Peter was emphasizing, these sheets on each of the things is really considerable, and those were not a part of the original draft we saw. They're added material.

DR. COGGINS: But at some point we're going to have to do that.

DR. HOWELL: Yes. I just don't think it will be done expeditiously.

DR. HAWKINS: I'm not a lawyer or anything, but I'm just looking over the minutes. Basically what we said, we've already approved the report, okay? So if we've approved the report, we can send it right now, and that's what we said in the minutes, and I think we're trying to figure out how to change our minds or

something. According to what we said, we've already approved the report, so why can't it go on to the Secretary right now?

DR. HOWELL: I think that was Jennifer's near-motion.

DR. HAWKINS: Exactly, although I guess my point is I don't think a new motion has to be made to send it. I think we should send it and then we should decide what's going to happen now after he gets it.

DR. HOWELL: I hear what you're saying, but I think that in view of the fact of the ambiguity that exists in the old minutes, it would be very nice to come forth with some unambiguous recommendations that we vote on so we don't have this discussion at the next meeting, because our patience may become weary. (Laughter.)

DR. TELFAIR: Well, mine is just a recommendation for Jennifer's third point, which is when the committee's and public comments are made, there clearly is going to be a distinction made between what the committee said and what the public said, that we go with the first part of this, which is the first point, which is you review that as well as a third point, and then forward that, instead of this committee having to rereview it yet once again or try to synthesize it once again. I'm just recommending how to resolve that problem at the end.

DR. HOWELL: Derek?

MR. ROBERTSON: I think one point that was raised last time that we probably have forgotten, but the point was made that the report is not this committee's report. We didn't generate the report. It was generated in a whole other system. Now, we do have some overlap. So I think that was one of the reasons that people eventually agreed to say, well, okay, let's go ahead and send it. So I think it would be appropriate to send it the way it is, and then we would still comment on it because it's an important report, and as Dr. van Dyck said, we would assume that the Secretary would want to know the input of this advisory committee based on the report.
So we are commenting on a report that's another body of people put a lot of time and effort into it and sent it on to HRSA, as they should have. They did a final report to HRSA. So the report goes, and I think in essence your letter was going to say that we think it's a really important report that was worked on, and here it is. The other option would be, then, when we get it we have comments on it as a committee. I think that's important, which would go along with anybody else's public comments.

DR. HOWELL: Coleen?

DR. BOYLE: I can get behind either forwarding the report as it is under your signature or the other motion, which was to have us review it and include our comments along with the report going forward. But I guess my one concern was that we get together as a committee and that we come to some consensus, and that it's a unanimous consensus about our recommendations on the report. That would be my one recommendation.

DR. HOWELL: It sounds like you supported Jennifer's near-motion, I believe. One of your commentaries suggested that it would be consistent with what I believe Jennifer was saying. Is that not correct?

DR. EDWARDS: I think the one thing I hear that's different from what Coleen said is that -- what I heard Jennifer saying is that we would individually send our comments in, and what I heard Coleen say is that it's actually the committee coming together --

DR. HOWELL: No. This is very important, because I didn't hear her say that.

DR. HOWSE: Piero made a very good suggestion through hand signals, which was why don't we take each part of this motion and make three motions and see how many of them we can resolve? Then we'll work on the ones that we don't like.

DR. HOWELL: So you have the first motion.

DR. HOWSE: So the first would be that the report be sent under Chairman Howell's signature to the Secretary of HHS.

DR. HOWELL: Is there a second for that motion?

DR. EDWARDS: Second.

DR. BECKER: Is the operative word there, Jennifer, "immediately"?

DR. HOWSE: Immediately.

DR. BECKER: Or upon availability, immediately upon availability?

DR. HOWSE: I think immediately. I mean, that was our feeling in September. I can only imagine that's grown more intense.

DR. BECKER: Okay. I'm comfortable. I just wanted to clarify.

DR. HOWELL: So you want to state that again so there will be no ambiguity in the record?

DR. HOWSE: I'd like to propose a motion that this report be sent immediately to the Secretary of HHS under Chairman Howell's signature on behalf of the committee.

DR. HOWELL: And that was seconded by Steve, and you still second that.
Is there any further discussion on that particular motion?

DR. DOUGHERTY: It's just a question for Peter. Can he release the report to the committee, since the Department hasn't given permission to release it publicly? I mean, immediately --

DR. VAN DYCK: I'm not sure. I'll have to check. But "immediate" to me means as immediate as I can possibly do it following the regulations and law.

DR. DOUGHERTY: There was this point about immediate versus upon availability, and that was what I was trying to get at. If that's the meaning of "immediate," that's fine.

DR. HOWELL: So the immediate as referred to in your motion means as soon as he can do it under the HRSA regulations, which he anticipates will be very soon.

DR. HOWSE: Right, to release the report under your signature to the Secretary.

DR. RINALDO: Do the two things coincide?

DR. HOWELL: They will, but that's a different issue.

DR. HOWSE: So that's the first motion.

DR. HOWELL: Those favoring this motion, let's see a show of hands.
(Show of hands.)

DR. BOYLE: I'm sorry. I actually feel like I need it as a package deal to understand what I'm voting on here, not step by step, because I might not agree with the second step.

DR. HOWELL: So what do we want to do? We have a motion that's seconded.

DR. EDWARDS: (Inaudible.)

DR. HOWELL: Everyone favored that, I believe. Is that correct? It was a unanimous approval.

DR. HOWSE: There's a general intention on the first part of this.
The second part, that the report be immediately released into the public domain for comment.

DR. HOWELL: Second?

DR. HOWSE: For appropriate comment.

MR. ROBERTSON: I don't know if it's our role to direct that the report be released for comment. Shouldn't we say that it be done along with federal guidelines? We would recommend that HRSA release a report to the public in keeping with appropriate guidelines as soon as possible. You can only recommend it.

DR. HOWSE: Right. We recommend the report be released into the public domain as soon as possible for review and comment. How's that?

DR. HOWELL: Second to that?

PARTICIPANT: Second.

DR. HOWELL: Anybody disagree with that?
Those favoring that, raise your hands.
(Show of hands.)

DR. BOYLE: A vote on the entire package.

DR. HOWSE: We are. We're just getting intentionality and we're trying to wordcraft.

DR. HOWELL: Number three.

DR. HOWSE: Number three, I need a great deal of help on this one, that we transmit the comments of this committee and -- maybe that's it, that we transmit the comments of this committee, period.

DR. EDWARDS: The issue that Coleen raised is the question about whether they should be individual comments or a group commentary. I think that's the ambivalent thing here right now from the way yours reads.

DR. HOWSE: So wordcraft that the way you'd like it.

MR. ROBERTSON: It's recommended that the committee review the report and make recommendations within the same time frame as the public comment.

DR. HOWELL: Is that a motion, Derek?

MR. ROBERTSON: Yes.

DR. HOWELL: Is there a second for that motion?

DR. HAWKINS: I'll second it.

DR. HOWELL: Greg seconds it. Okay. So we can discuss that motion.

DR. BECKER: Rod, I still think Steve's point is on the table out of that recommendation. Will the committee's comments be committee comments, or will they be individual committee members'

comments over the same time period?

MR. ROBERTSON: I'm thinking it's the committee's comments. Individuals can always write individually, but I think it's the committee that has to make recommendations.

DR. BECKER: Can I suggest that that be clarified in your recommendation to make sure?

DR. HOWELL: Is there a general sense that it would be the committee, and then we can ask our person if he would be willing to modify that to be clear?

MR. ROBERTSON: I would be willing to do that.

DR. HOWELL: And you would agree with that, Greg?

DR. HAWKINS: Yes.

DR. HOWELL: Why don't you make it clear, then?

MR. ROBERTSON: Recommend that the committee would review the report and make comments as a committee within the same time frame as the public comment.

DR. HOWELL: I think that's very clear.
Greg, do you second that?

DR. HAWKINS: Yes.

DR. HOWELL: Those favoring that?

PARTICIPANT: Can we have a discussion on that?

DR. HOWELL: Oh, sure. Derek may kick you under the table, but of course.

DR. DOUGHERTY: I'm just thinking of perceptions and that to send the report now to the Secretary suggests that we've all read it. I'm not sure what the letter is going to say. Then to review it after we've sent it seems a little odd to me. At least in the last vote, we had all seen the report. It was in the binder. We knew what we were voting on, and I think we voted to send that particular draft, but we have faulty memories on that.
But it just seems to me that we could say to the Secretary that we're sending you this report -- like the language last time was we recommend the report to you, and we will get you our comments within 60 days, which to me seems a little odd to be recommending a report that we haven't seen yet. But I'm not saying that means we shouldn't send it right now. I'm saying that we should not actually make a -- we can vote to have everyone review it and make minor comments, but I'm not sure we should be making a big deal of sending our comments after we've recommended the report.

DR. RINALDO: I have to agree with you, because it doesn't seem to be the proper chronological sequence. One of the ambiguities of the whole discussion is about the perception of the reality of making changes to the report based on the comments, and I think if we all understand that it's a final version and you may love it, hate it, or so and so, have some comments, that can be sort of put in a reasonable context, I think it's a different story. But we have to be clear on the fact that it's final, because it's not a product of this committee. It's submitted to this committee.

DR. HOWELL: Derek?

MR. ROBERTSON: Couldn't we theoretically, if we make recommendations and the public makes recommendations, can't the Department take those comments and make changes to the report if they think those comments are significant? Let's say there's an error that somebody in the public points out

about something. Wouldn't you then change it?

DR. VAN DYCK: I'm not sure what the process would be in the Department. That would be up to the Department. My sense would be that the report is final and the report is the report, and the comments are appended to the report.

DR. RINALDO: So if it is final, then I think we can feel comfortable following the process outlined by Jennifer.

DR. DOUGHERTY: I think you're right, that rather than saying comments on the report, send recommendations for follow-up. You can send the report now and say we'll be back to you with our recommendations for things to follow up on that the Department might do to facilitate use of this report, or something like that, which is a logical sequence.

DR. RINALDO: Yes, but the point I would make is we might follow up with an analysis of the comments made on the report. So that is a different thing, rather than a revision of it. If we agree that we will do an analysis, there's always room for improvement. So we can go on, and perhaps we could reedit the Ten Commandments.

MR. ROBERTSON: I think the main point is that it's not our report. All we can do really is comment on it. They're presenting a report from HRSA. It's not our report. But we do recognize that the report is an important document, and we're saying yes, Mr. Secretary, you should have this report, and then we can comment on it.

DR. RINALDO: And we'll follow up with an analysis of the comments.

DR. HOWELL: Steve?

DR. EDWARDS: I wonder if we shouldn't drop step three and go to step four, and step four would be that when all the commentary is -- I think this is the point that Piero was getting to, that since we are recommending it to the Secretary, that we not comment again on what we're recommending. So drop step three, but then go to step four, which would be reviewing all the comments that come in and make additional recommendations based on the commentary that's come in.

DR. RINALDO: We may or may not.

DR. EDWARDS: Yes, but then we'll look at it again after.

DR. HOWELL: Duane?

DR. ALEXANDER: A lot of the resolution here depends on what goes in the cover letter, and what you say you're transmitting and what you're endorsing. Really, in this situation in which we are where we haven't seen this final document, it's hard to submit a cover letter that endorses everything in that document. What you're really doing is transmitting a report for the Secretary's consideration that was prepared by a contractor to HRSA and calling it to the Secretary's attention and urging that he take action on it, and that that action include putting it out for public comment and responding to that comment and implementing policies as quickly as possible, and offering this committee's, which reports to the Secretary, assistance in responding to that and offering guidance to the Secretary on implementation of any policies that would come forth from that.

I mean, the report is not an action document so much itself. It's a status report. It's suggestions for things that ought to be included in screening. It's suggestions for things that need to be taken into consideration as you move to expand screening. But in itself, it's not an implementation document for the Department. What the Department will do with this, ideally what we would hope they would do, would be take this document into consideration as a framework for what actions would be taken to expand, to modify, to whatever newborn screening.

So what we would do in this cover letter is commend this to the Secretary's attention, we think it's a very important document, and urge the Secretary to put it out for public comment as quickly as possible and offer our services in commenting on it and advising the Secretary on converting this document into policy. That's what a cover letter would do in this case. Then when the Secretary follows your good advice and puts it out for public comment, you are participants in that process of making the public comments and suggesting what we like and what we don't like in the report and how that might be translated into policy, into practice, into operations by the Department.

DR. RINALDO: I agree. However, as perhaps one of the few people who is painfully knowledgeable about every single version, the truth is that the version this committee reviewed in September and the final version has a lot of whistles and additions and gaps filled, but the conclusions and the recommendations, with the notable exception of dropping one condition based on the comments that really were stirred by the meeting in September, are identical. They are identical. There is a lot more material that really was driven in great part by the comments of the people.
Again, not being involved with the expert panel and seeing it for the first time, using very valid motives and points, said, well, what about this? I remember sitting next to Dr. Dougherty who said, well, this is or is not there. Well, it was an omission. It wasn't that it wasn't done. It wasn't included in the report. But the conclusions and the recommendations are identical.
So I know Dr. Howell will monitor my temperature. That's probably why I'm sitting next to him.
(Laughter.)

DR. RINALDO: But I remember getting pretty fired up when I said, okay, you are 99 percent. How much effort and time do you want to put to go to 99.5 when there is a more than likely chance that nothing will change? There will be more transparent methodology, more comprehensive data, but conclusions and recommendations are identical. I was thinking that perhaps Dr. Telfair is the only person in America who hasn't seen the report yet, so probably he's the one who should get a chance to look at it.

DR. TELFAIR: I just got here, so don't volunteer me. Thanks.
I'm kidding.

DR. RINALDO: But I want to stress the point: Conclusions and recommendations are identical of what was voted upon and what was discussed.

DR. HOWELL: I think we've had actually, surprisingly, pretty good consensus about how we go, and I think the question is that we had a motion and a second about one issue, and it's been in the discussion suggested that that one be dropped and that a fourth one be added and so forth. What's been your response to that motion or seconders?

DR. EDWARDS: The only thing that I object to, if Derek will give me the floor, is that the committee should review it again during that 60-day commentary period and offer its comments. I would prefer seeing the committee review all of the comments, and then at some point later maybe offer some recommendations if we consider it appropriate. I think it's appropriate for the committee to look at it again. So I would suggest we drop that committee review because it sounds like we're saying we recommend this to you but we want to review it, and I don't think we should do that.

DR. HOWELL: Derek, your response?

MR. ROBERTSON: Well, I guess I agree with Dr. Alexander's comment that we're not necessarily recommending the report. What we're saying is that here's a very important document that was developed by a contractor and given to HRSA, and we want you to see it and we want you to review it and we want you to get it out to the public.

DR. RINALDO: To see, look, this is great, but just being noncommittal I think would be quite a concerning position.

MR. ROBERTSON: But we did recommend the report.

DR. HOWELL: Yes.

MR. ROBERTSON: Then I will yield to Dr. Edwards.

DR. HOWELL: And with the second, Greg, would you agree?

DR. LLOYD-PURYEAR: Could we read the motion?

DR. HOWELL: We have a new motion, because they've agreed to modify it.

DR. LLOYD-PURYEAR: But Steve's motion, can we read it?

DR. HOWELL: But we're trying to get rid of the other one.
Steve, now you want to come up with a new motion?

DR. EDWARDS: Well, somebody else can state this better, but basically all I wanted to say is that we're not through with this once it's over. We're continuing as a body. We've got meetings scheduled through the rest of this year, and I think it would be irresponsible of us to say that we've sent it to the Secretary and forget about it. I think we should review the comments that the public have made and see if we want to modify any of our suggestions to the Secretary. It's the Secretary's report now once we send it. It's not our report anymore. But we're still an advisory body.
So that's all I would suggest, that as a part of our continuing meeting that we review the commentaries and then make recommendations if we feel that they're justified.

DR. HOWELL: So that's the motion. Do we have a second for that?

DR. ALEXANDER: We have to be a little bit careful of how much authority we have as an advisory committee. The process here is that the Department would seek comments on this report if they go along with the request that comes from HRSA as an agency, and chances are that they would. We are part of that comment process. We probably have very special standing since we're the Secretary's advisory committee on this, and the cover letter ought to indicate that we would plan to provide comments to him and advice to him in our capacity as his advisory committee on this report during the public comment process, and we're fully entitled to do that.
We have no standing on our own to insist that we get public comments from everybody else. What we could do in the cover letter is ask the Secretary to provide us an opportunity as his advisory committee to provide additional advice to him and his Department by looking at the other public comments too if he were willing to make them available to us. But we would not ordinarily be party to those other public comments. What happens is that those public comments would go back to the originating agency, in this instance probably HRSA, and HRSA then would convene representatives from other relevant agencies of the Department to review those public comments and prepare a response to those, which in many instances, and probably here, would be eventually published.
We could request in our cover letter to have an opportunity to participate in that process of review of the public comments or at some stage, but that's the entree that we as a committee, a federal advisory committee, would have.

DR. HOWELL: Would you like to modify your motion?

DR. EDWARDS: That would be fine.
The only thing that Duane said, though, that is getting us back to Derek's, what Duane said is that if we comment during the commentary period, and I thought that's what we just elected not to do.

DR. VAN DYCK: I also would hate to see the committee give up its opportunity and, really, strength to not comment during the comment period. I think the Secretary would request, would want comments from this committee during the review period.

DR. BOYLE: I don't think that was dropped.

DR. VAN DYCK: We dropped the third piece and went to the fourth, and the third piece was commenting during the comment period. I hate to see us do one or the other.

DR. HOWELL: Would you like to add the commentary period?

DR. EDWARDS: Derek's got the wording. If that's what the committee wants to do, Derek made the recommendation. The committee can comment during the commentary period. But I would like to leave the fourth standing alone, the recommendation that we include in our letter a suggestion to the Secretary that we would like to look at the commentary and, if we felt necessary, to be free to advise him on it.

DR. RINALDO: I have to agree. I think this is perhaps the first suggestion where we don't have a pretty obvious chronological discrepancy. So we would like to see what people besides the committee have to say. In this way, the chronology I think is rational and credible. So I think it's an excellent idea.

DR. HOWELL: Okay. We have, then, three and four. Is that correct?

MR. ROBERTSON: So let me just clarify, then. What we're saying is that it's okay to send the report, recommend the report to the Secretary and also make comments as a committee.

DR. EDWARDS: That's what he said.

DR. DOUGHERTY: I would add the word "additional" recommendations in the phrasing. What I wrote down is review the comments made by the public and the committee would make additional recommendations if we feel they're justified.

DR. RINALDO: May make.

DR. DOUGHERTY: May make, but I would use the word "additional" because what we're recommending to the Secretary is the report. So to just say recommendations makes it a little fuzzy in the chronology.

DR. HOWSE: I'd like to suggest that we ask that a draft of this transmittal letter be prepared so that we can look at it tomorrow, because really we've got so much riding now on the excellent suggestions that have been made about what goes into this transmittal letter. All four elements are included. If we had the draft of that letter to look at, I think it might aid us in our deliberations, and then we could simply vote on the letter and it would contain within it these four elements that we've been really working hard to try to refine to everyone's satisfaction. Is that a possible thing to do, Peter and Michele? Would that be okay with everyone? Then we have text in front of us, and I think we've progressed maybe about as far as we can verbally at this point.

DR. HOWELL: We still have motions that have been discussed that have not been finalized nor voted on. Do you want to talk about number 3?

DR. HOWSE: Let's table the motions until we see the text.

DR. HOWELL: You want to table all the motions until you see the text?

DR. HOWSE: Yes, and then we can pass a motion that says we approve this letter, and it will have the elements in it.

DR. RINALDO: Could we use the computer still hooked up, so using the word processing just outline it? Because I think hearing them, but if we see them on the screen perhaps it would be easier for us. Can we quickly do it?

DR. LLOYD-PURYEAR: You've already voted on two motions unanimously.

DR. RINALDO: We still have some open --

DR. HOWSE: That was a test.

DR. EDWARDS: I think that's right, because what we had said is we wanted to put this into a -- well, we did vote on them, but we were seeing if there was consensus among us, and then the idea was to put those into a package, and what we haven't done is completed the package.

DR. RINALDO: I would like to see the package.

DR. HOWELL: I have some anxiety about tabling all these motions and coming up tomorrow with a letter, because the inauguration is Thursday and we may have to check into the hotel or something until then to work on this. But anyway, we'll go ahead and do that, but I think that we will have to be much more brisk in our discussions than we might have been today.

DR. HOWSE: Can we see that text tomorrow morning so that we can go ahead and get this business dispatched tomorrow morning? Just to follow along on your point. I'll be happy to volunteer to draft the letter if that will help everybody's minds.

DR. LLOYD-PURYEAR: The letter is already there.

DR. HOWSE: Right. Well, then, let's do that and it can be amended according to the discussion.

DR. LLOYD-PURYEAR: If the committee would like to write its motions down, that would be helpful.

DR. HOWSE: Well, the motions really need to be reflected in the letter, I think is what people are saying, that the sentiments that we've been discussing and the specifics that we've been discussing we want to have as part of the letter of transmittal because it really makes our position quite plain and clear. So if the letter as it sits now could be amended to reflect what we've discussed, the four elements we've discussed this afternoon, we could look at that in draft form tomorrow morning and then vote on the letter, and that will contain the elements of the motions that we've been trying to craft verbally this afternoon.

DR. VAN DYCK: In all due respect, I'm a little sensitive to having staff draft a letter which tries to incorporate the sense of the committee. I'm a little uncomfortable doing that. I would have no problem with the committee drafting a letter to do that, but I'm a little sensitive about assigning that to staff when the sense of the committee is unclear.

DR. EDWARDS: I think the sense of the committee is very clear except for one item, and that is item number 3. The decision on number 3 is does this group want to comment on the report that we send to the Secretary during the period that it's open for public commentary? I think that's the only thing that we haven't made a clear decision on, and Derek made a motion that we do that. I was kind of feeling like that we shouldn't, but I have no great objection to it. But I think if we make that decision, then we've decided the issue.
Also, I think staff all the time consolidates the discussion that they've heard, and it won't be staff that's approving the letter, it's going to be this committee that approves the letter.

DR. VAN DYCK: Well, I think if we can get a sense of that third item, then it becomes more clear.
MR. ROBERTSON: I guess, like I said, I think maybe we just need to get a sense -- the same thing happened the last meeting. It all stems from -- I think there are some on the committee not being entirely comfortable sending something forward that they haven't seen. I mean, that's where it all stems from. So we just have to decide and then we can go back to number 3.
I think the question is two things. One is do we want to recommend the report in this state to the Secretary? That's either yes or no. If we say yes to that, then the second question that follows is do we still, after making that recommendation, want an opportunity to comment as a committee during the comment period? I think some people are saying, well, if we've made the recommendation initially, then

how can we go back and make more recommendations during the comment period? That's what the whole debate is about. So I think we just need to answer those questions.

The first question is are we recommending the report to the Secretary?

DR. EDWARDS: Item number 1 that we voted on, Jennifer said yes.

MR. ROBERTSON: So then the second question is do we want to make comments during the comment period as a committee?

DR. HOWSE: Why don't we say the committee will consider its recommendations on the report during the public comment period.

MR. ROBERTSON: What I wanted to do is just get something really simple. It's a yes or not issue. So let's do that, and then wordsmith it. We've answered one question. We want to recommend it. Fine. The second question which gets to this number 3 is do we want to make recommendations as a committee during the comment period?

DR. RINALDO: But that really begs the question why later?

MR. ROBERTSON: Then the answer to that would be no. I'm saying let's answer it. Let's not discuss it, let's answer the question. You would probably say no, we don't need to.

DR. RINALDO: Yes.

MR. ROBERTSON: Then that's what we need to get from the committee.

DR. RINALDO: But I just want to say one more time, I think that claiming ignorance of the content of the report is a questionable point.

MR. ROBERTSON: I know, Piero. But what I'm saying is let's get past that first. That's why the discussion is going round and round. We either want to make the recommendations or not. Do we want to comment as a committee, or do people think it's not necessary because we've already recommended it? I think you would probably fall on the side of saying no, we've already done it. What does the rest of the committee feel? Do we want to make recommendations during the comment period, yes or no? Then we can kind of get rid of number 3 or keep number 3 and move from there.

DR. TELFAIR: It's yes or no. I think the Chair should call for yes or no.
MR. ROBERTSON: That's my suggestion, Mr. Chairman.

DR. HOWELL: Any further discussion before we say yes or no? Does the committee, during the public comment period, want to comment on the report as a committee?

DR. BECKER: Derek used the word "recommend." You used the word "comment." There are some distinctions I think being made around the table between the two. So we have to be very, very clear what we're answering yes or no to. Are you asking should we comment?

MR. ROBERTSON: Well, I think "comment" is a more general term because it recommendation is a form of a comment. So you could use the more general term "comment."

DR. BECKER: I think there's a connotation to the word "recommend" since we're already recommending the report to the Secretary, and it seems a little redundant to make additional recommendations.

DR. HOWELL: Well, that's why I used "comment."

DR. HOWSE: Ask do we want to comment during the comment period. Why would we not?

DR. BECKER: I would vote yes for that.

DR. HOWELL: Well, those favoring commenting on the report during the public comment period as a committee, raise your hands.
(Show of hands.)

DR. HOWELL: Those that would not want to comment?
(No response.)

DR. HOWELL: That's clear, I believe.

DR. HOWSE: So is there a recommendation, number 3?

DR. HOWELL: That was yes, during the public comment period.

DR. HOWSE: So the recommendation is the committee will comment on the report during the public comment period.

DR. RINALDO: And it will be up to the committee to make sure --

DR. HOWSE: The committee will review the report and make comments as a committee during the same public comment period. The committee will comment on the report during the public review period.

DR. RINALDO: And it will be the responsibility of the committee to make sure we avoid conflict and really confusion between the recommendations and the comments.

DR. HOWSE: We'll figure it out.
And number 4, the committee requests access to other public comments for its review and consideration.

DR. HOWELL: Any further? Would folks agree with that? We now are not talking about motions but we're talking about the content of this letter. Is that right?

DR. TELFAIR: That's correct.

DR. HOWELL: I see nodding of heads. I think that's what we were talking about.

DR. HOWSE: Well, we can have it both ways. We can make these motions, and then we can ask to see the text of the letter, which I think we ought to look at before it goes. We can ask to see the text of the letter to be sure that our motions are appropriately considered.

DR. HOWELL: Well, there are four points that we have discussed as motions in and out and so forth that we will try to get in the letter. The letter has been drafted and the letter does not contain much of this stuff at the current time, which would not surprise you.

MR. ROBERTSON: The only problem I have with circulating the letter, Jennifer, is that it's so many of us, and if we're going to get into wordsmithing -- maybe the chairman is going to review it. He knows the sentiments, and I think we should just go with the fact that he's going to include what we've asked him to include, because I think it's going to get into another round of this.

DR. HOWSE: I can be persuaded. But then I do think we need to make these motions so we have a record in the minutes about what the desire of the committee is.

DR. BOYLE: I also would like the minutes -- I was going to say this initially when you asked us to approve the minutes. I found them to be more of a summary than an actual statement of it, and I don't know whether -- since it was so confusing last time for me personally, I read the last part of it on the plane

coming up, and I thought, well, I'm not even sure this reflects what happened. So I feel like maybe our minutes, especially the motions, need to be in quotes somehow so we actually have a reflection of what we voted on, maybe not all the other nuances that we just talked about, but that wasn't well described in the minutes.

DR. HOWELL: I think we've had an adequate amount of discussion about this, but it seems to me that it's a bit unusual to have an entire committee wordsmith a letter. Unless there is some compelling reason to do that, I think we should not do that.

DR. HOWSE: It was just an attempt to be a proxy for the fact that none of us have seen the final report. A few of us have.

DR. HOWELL: On the other hand, the four recommendations that have been discussed extensively I think are clear now, and in view of the fact that we're not going to circulate the letter, I think we should, God help us, go back and go through the motions again, but that should be really quite brisk.
Jennifer, why don't you do the first two since they were in your court?

DR. HOWSE: That we would --

MR. ROBERTSON: Didn't Michele --

DR. HOWELL: Michele can read them.

DR. LLOYD-PURYEAR: Recommend that the report is sent to the Secretary under Chairman Howell's signature immediately on behalf of the committee.

DR. RINALDO: Sent or recommended?

DR. LLOYD-PURYEAR: Sent. Recommend that the report be sent to the Secretary under Chairman Howell's signature immediately on behalf of the committee. That was unanimous.
Recommend that the report be released as soon as possible into the public domain.

DR. HOWELL: Number 3.

DR. LLOYD-PURYEAR: I've got to find number 3. Recommend that the committee review, report, and make comments as a committee during the same public comment period.

DR. HOWELL: That's it.
Number 4.

DR. LLOYD-PURYEAR: Well, number 4 was never in a recommendation, because it's the committee requesting an opportunity to review the public comments and provide advice to the Secretary. So that's a request. That was not ever put in the form of a recommendation.

DR. HOWELL: Would someone like to make that as a motion?

DR. EDWARDS: I think we're recommending this report, not just sending the report.

DR. LLOYD-PURYEAR: The committee last time -- and if people want to see all 800 pages of the transcripts, I'll be glad to email them to them. The committee last time recommended the report.

DR. BECKER: Yes. Michele, I think the question that is being raised is when on that first letter motion, what we're talking about here, where we're sending the report to the Secretary, it's under the auspices of what we've already approved that we're accepting it and recommending it, and that's the manner in which it's going to be sent to the Secretary. That really wasn't said but I think that's the concern that folks just wanted to verbalize. That's how you're understanding it? I'm perfectly fine with that.

DR. EDWARDS: But initially what you wrote and said to us does not say recommend, it just says send.

DR. LLOYD-PURYEAR: Because that wasn't the motion. I'd go back to notes, because it was under the assumption with the understanding that the report had already been recommended and we were sending what was considered the final report to the Secretary under Chairman Howell's signature. But the report had already been recommended.

DR. EDWARDS: But the Secretary doesn't know that.

DR. HOWELL: No, he doesn't.

DR. EDWARDS: No, because it hadn't gone.
I think my point is Bill's point, I think, that it should clearly state in there that we are recommending this report, not just that we're recommending that it be sent but we're recommending the report.

DR. LLOYD-PURYEAR: You mean the letter.

DR. EDWARDS: The letter should say we're recommending --

DR. LLOYD-PURYEAR: Yes, it does. This was a recommendation. I didn't read the letter. I wasn't reading the letter, I was reading your recommendation.

DR. EDWARDS: I'm saying that our recommendation says that we're recommending it to the Secretary.

DR. HOWELL: The letter that goes to the Secretary you say should say we are recommending this report. That's the point that you're making. Is that correct?
Is there general agreement to that?

DR. BOYLE: I just have to say again, as I said last time, I felt uncomfortable recommending a report and then commenting on it. But I can get behind the fact that I would like the committee to comment in a unanimous way about the report, and since that's in that motion now, I feel like I can go for it.

DR. HOWELL: So is there a unanimous sense, then, that the letter will say we're recommending this, and we will have all the other stuff and so forth? I will try to capture all the wisdom of this group, which will be difficult because it's been so extensive.
Is there anything else to say about this report? I hope not.
(Laughter.)

DR. HAWKINS: Are we going to vote on the fourth thing about wanting to see, or is that just going to be in the letter?

DR. HOWELL: That's going to be in the letter.
Would someone like to make a motion about the final fourth thing so it will be a matter of record?

DR. HOWSE: I'm sorry.

DR. HOWELL: The fourth thing. Do you want to read what you wrote down? And we can make it as a motion.

DR. HOWSE: That the committee request access to all public comment for its review and consideration.

DR. HOWELL: Did you get that?

DR. LLOYD-PURYEAR: I had that the committee was requesting access to the public comments but an opportunity to provide advice to the Secretary on those public comments.

DR. HOWELL: That's a motion, then. Is that acceptable?

PARTICIPANT: So moved.

DR. HOWELL: And you accept that as a second?

PARTICIPANT: Second.

DR. HOWELL: Those in favor?
(Show of hands.)

DR. HOWELL: Unanimous. Let's move ahead.
It's 5 o'clock, Peter, and we've not heard your major talk this afternoon.

DR. VAN DYCK: It's not a major talk. I was just going to run through items that we felt the committee should think of. I was going to run through quickly all of the items that we collected that we feel are potential elements for the committee to consider as they divide into subcommittees. If the committee is willing to sit for five or ten minutes, I think we could do that.

DR. HOWELL: Thank you. That would be wonderful.

DR. HOWSE: Peter, we've waited years for this report, so we're willing to wait to hear what you have to say to help guide our work.

MR. ROBERTSON: Mr. Chairman, with your permission, I'd like to be excused.

DR. HOWELL: We will miss you, Derek. Thank you.

DR. VAN DYCK: I usually stand when I talk, and particularly at this hour, but there's not a traveling mike, so I'll sit.

We in Maternal and Child Health, as we include the services we offer in Maternal and Child Health, we try to find a way to best think about it, and we try to define it by the pyramid. In this pyramid we put direct health care services at the top, basic health services, services in a physician's office or a clinical office. It's clear what those are, direct clinical services.

The second layer of the pyramid is enabling services, those kinds of services that enable people to get into those direct care services, transportation, translation, outreach, case management, care coordination, those kinds of things.

The third layer of the pyramid are our population-based services, newborn screening, lead screening, immunization, nutrition, counseling, things that are delivered to an entire segment of the population or an entire population group within the population, or to all populations.

The bottom of the pyramid are infrastructure services, evaluation, planning, policy, standards, guidance, quality, needs assessment.
They're laid out in this order because we feel that the top of the pyramid can't work well without a good bottom and structure in infrastructure and population-based services. So as we thought about outlining the elements we feel were important for the committee to consider, we did it using this pyramid.

Now, if you turn to Tab -- I think it's 9, and you turn past my slides, there is an outline that has the slides. Is it Tab 11? Turn past the slides and there should be an outline, and I'm just going to go through this outline. The pieces that are bolded are those elements the committee has already heard a discussion

about or will hear a discussion about, or has today or tomorrow. So there are a few items bolded. So you might want to make your notes on that outline as we talk, and I'm going to go through these very quickly.

Infrastructure services, policy development is one of the main areas. So the whole area of laboratory standards, cutoff values, case definitions, testing strategies, turn-around time, nomenclature, and report forms. These are all issues within the laboratory standard piece of policy development we felt were important, and none have been discussed yet by the committee.

To continue, policy development, policies for the use and storage of residual blood spots, follow-up protocols once a case is identified, what is a standardized protocol for follow-up, what should be the appropriate panel recommendation for screening conditions -- that's bolded because we've had a discussion about that, which doesn't mean we can't discuss it again or continue it, but at least it's been discussed formally before the panel -- and informed decision-making around consent or dissent.
Then there's informatics infrastructure, tracking and surveillance. We've talked about tracking and surveillance briefly, definitions and principles of core functions, and defining a data dictionary. We also have discussed or will discuss short- and long-term tracking capabilities, the knowledge base, and how do we evaluate that.
Further under infrastructure, integrated information systems development. You heard a discussion of that this afternoon, so it's bolded. Integration of the public health and personal health services data and the records, provider access to newborn screening results, how does this all fit into the medical home concept, and how do we deal with the privacy and confidentiality issues. The linkage of clinically useful data using a child health profile concept, and working on continuous quality improvement are all an important part of the integrated information systems development.

Then training, training on genetics, diseases, treatments and technologies, the training of health care professionals, both primary and subspecialty care professionals, public health professionals, laboratories are all important training components to make this whole system work.

Continuing under infrastructure, research, the continuing research to develop new therapies, and developing new testing platforms and strategies.
Evaluation. We heard last meeting about cost effectiveness evaluation from a couple of states and one university. Pilot testing of a technology, clinical validity and utility, and health outcomes evaluation.

Then the quality assurance piece, policy development for laboratories around data standards, case definitions, cutoff definitions, and the policy development for programs, a broad area under quality assurance.

Continuing under infrastructure, distance communication, what can we learn from this, how do we incorporate this into the newborn system, issues related to reimbursement, legal issues, and the infrastructure, setting up the infrastructure for this distance communication.

Then a major topic under financing of newborn screening system infrastructure, we talked about this at the last meeting briefly.
So those are the issues around infrastructure that have arisen in our discussions, arisen in the discussions that the committee has had and arisen from input into us from various sources.

Then there's some population-based services. Education, again education around health care and public health, primary health care and public health professionals, hospital, obstetric and pediatric health care, as well as family practice and the other primary care specialists, education of the general public, and education of parents.
Moving up the pyramid to enabling services. Access to therapies and supplemental services and therapies such as OT/PT, early intervention, Title V is Maternal and Child Health, and the array of maternal and child health services such as multidisciplinary teams and care, and how do we incorporate the concept of medical home and children with special health care needs.

Then as we talk about these therapies and supplemental therapies, how do we deal with reimbursement or what recommendations do we make around reimbursement, and we don't want to forget nutritional support as well.

Then family support activities, family support advocacy activities, as well as educational activities for families.

Then in direct services, issues that have arisen include genetic counseling, and then the issues around treatment, subspecialty, access to subspecialists, availability of subspecialists, distribution of subspecialists, the same for the nutrition services, and the same for other necessary treatment and services for comprehensive care.
And that's just a quick run through of the items we feel should be considered, at least these items should be considered as we think about dividing into subcommittees, and I'm not suggesting that they be divided as infrastructure, population-based, enabling. That was just a mechanism for us to think about it. But I think if you look at this list and make some notes, there may be some more obvious ways that there can be a division among a subcommittee infrastructure.
Any comments? I know it's late and everybody is interested in leaving.

DR. HOWELL: Amy?

DR. BROWER: I just have a quick question where we talked about the report from ACMG recommends a certain panel. But as new tests become available and clinical utility gets proven, where do we address the pipeline of new tests and new conditions? Is that under panel of screening conditions or testing strategies?

DR. VAN DYCK: Yes, that's there, and you probably should add a note there to remind yourself that that's part of that area. The report does make recommendations about a standard way to include new tests as they become available.

DR. HOWELL: Bill?

DR. BECKER: Peter, a question for you. In the pyramid structure, you have under enabling services health education and family support services. But in your outline, you have education of the general public and parents listed under population-based services. I was wondering what the distinction would be there. Could that just as easily be education under enabling services?

DR. VAN DYCK: I don't think it's so important where it is. I think the concept is, under population-based services it's educational services universally to a population, to all pregnant women or to all hospitals. Educational material developed for enabling purposes would be more to enable people to access care or the reasons for accessing care, or their ability to interact in a physician's office or the questions to ask. I mean, it's a mild difference, but in our conceptual structure that's the difference.

DR. BECKER: Okay, that's what I was looking for is clarification. Education is unbelievably important, and I was just curious about the distinction, that's all.

DR. RINALDO: Following Bill, actually I was thinking along similar lines. I see the educational professionals actually being very much part of the infrastructure building, because we certainly recognize shortages. So I would say that some of these are so important that they should be spread throughout the entire pyramid. They might be placed in one context, but they certainly belong in the others too.

DR. VAN DYCK: Right. Again, this is just a construct to help us kind of place things as we think about them.

DR. HOWELL: Jennifer?

DR. HOWSE: Peter, I think this is a very useful formulation, and as we increasingly turn out eye as a committee towards the very real implementation issues that are occurring at the state level, as I looked at the list and considered the pyramid, to me there were three areas that I thought were really plugged into implementation but also comprehended your list.

I don't know, maybe this is a subcommittee structure suggestion and we can take it up tomorrow, but one is scope and quality of the newborn screening program, because scope brings in a lot of the discussions about the panel, the cutoff, the tracking, sort of keeping track of what's out there, and then quality issues we had a lot of discussion about. So scope and quality seemed to pull a number of these categories.
Second, financing. Financing of the program in addition to what you said about infrastructure, but also just the basic financing of the program, how much is it costing now to do it. We've looked at a lot of information about fee structures and where does the money come from, and what's state and federal and private. So financing for the program I think is a very interesting implementation question and something that's on the mind particularly of a lot of state legislatures, getting good financing information, including more cost/benefit studies, like what George Cunningham did in California. The state legislature there really put a lot of credibility into that.
The third area, just to echo what Piero and some others have said, is that the category of education and training for professionals, parents and the public is a very, very, very important subject I think we've all been trying to grapple with.
So maybe this is just another way to slice and dice the issues, but I was trying to look at your list, Peter, in the pyramid and contemplate it in terms of where is the action with respect to implementation and how can the committee with all of its resources and wisdom and knowledge be the most supportive at the state level. So just some observations at the end of a very productive but nevertheless long day.

DR. HOWELL: Any further comments?
(No response.)

DR. HOWELL: Peter, I think this is a very helpful presentation. Again, I think that our next step is to look very critically at some subcommittees that can really move areas along, and this is a very excellent document to have and proceed on.
Let me thank the committee for your very hard work. I think we spent a long time on the ACMG report today, in great detail, but I think that's a very, very important report that underlines so much of newborn screening, and I think it's very helpful to rehash some of the questions so we're all clear. In the final analysis, I think we're in fairly full agreement on where we should go with that and so forth.
So thank you very much, and we'll see you at the crack of dawn.
(Whereupon, at 5:20 p.m., the meeting was recessed, to reconvene at 8:30 a.m. on Friday, January 14, 2005.)

U.S. DEPARTMENT OF HEALTH AND HUMAN SERVICES

ADVISORY COMMITTEE ON HERITABLE DISORDERS

AND GENETIC DISEASES IN NEWBORNS AND CHILDREN

Third Meeting

Friday,

January 14, 2005

Rotunda Room, 8th Floor

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1300 Pennsylvania Avenue, N.W.

Washington, D.C.

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C O N T E N T S

P R O C E E D I N G S (8:40 a.m.)

DR. HOWELL: Good morning. Welcome to this very rainy morning. I congratulate you for weathering all of the storms to get here. We will try to get underway in a relatively speedy way.

We have two important presentations to begin the morning. First, we are going to hear again from Brad Therrell, who is Director of the National Newborn Screening and Genetics Resource Center. He is going to be talking about the National Newborn Screening Data Initiatives.

Brad?

DR. THERRELL: Thanks, Dr. Howell.

This was already sort of set up yesterday by a question from Dr. Becker, so hopefully we'll answer that as we go through. What I want to do first is to go back and give you a little bit of history of how we got started into national data collection with HRSA support, where we have gone over the years, where we are now, and where we are going, we hope.

So to begin with, the Council of Regional Networks for Genetic Services, which was a HRSA-funded initiative that began back in the late '80s, had a couple of issues that they wanted to address, one of which was data. So there was a Data and Evaluation Committee which took on the job of evaluating genetics data, genetics service data, and newborn screening data.

So in 1988, they published their first report. I have given you a copy of that 1988 report, because I want you to look at the difference in 1988 and then 1998 as we go through here. So 1988 was the first report, 1989/1990, all these reports were similar in size to the one you have in your hands. They were done by the Data and Evaluation Committee.

Now, the problem with that report was that it defined things a certain way and asked for data a certain way. Lots of programs didn't have data collected that way, so they just didn't report. So that committee transitioned the data to the Newborn Screening Committee, which then reevaluated the questions that were being asked, and put out a little different format for the report.

So the first report of that committee was in 1991. At that point, there was also a data collection effort being mounted by the state and territorial public health laboratory directors. So we went to them and agreed to work together on these data, and to have this report that was already being collected by CORN serve to collect the data that ASTPHLD wanted. So at that point, we then began to cooperate with ASTPHLD, which is now known as APHL, or Association of Public Health Laboratories.

This is the validation and collection procedure that we went through. We had the questionnaire that asked for the data defined and developed by the Newborn Screening Committee. This committee had representation from all of the regions of the country, and had pretty much even distribution of laboratory and follow-up persons, so they developed a questionnaire. The questionnaire was then sent out and completed in the programs by laboratory and follow- up, and then it was approved internally before it was submitted.

We found that if you asked these questions of either laboratory or follow-up without getting the other's approval, you got different answers. So we then went to this approval process where whoever filled it out had to get the approval of the other group. I don't know why two and three didn't show up, but we have four.

This questionnaire was then submitted to a central collector, which in all cases since 1991 has been my group. We worked under subcontracts with CORN initially, and now we have this as a project of

the National Newborn Screening and Genetics Resource Center. So we have been involved in it since 1991.

Data is extracted from those questionnaires and put into tables. The tables were reviewed by the committee or the subcommittee and sent out to the programs to be reviewed, and then it was published. Periodically the Newborn Screening Committee would take a look at the questions and think about were these the right questions to be asking for the programs, were these the right data they wanted to collect, modified if necessary, and then move forward.

Actually over the years it was modified very little. So we were able to take a 10-year look at the data from 1990 to 2000 and collect some 10-year data for the country, which I have given this committee before.

So in 1991, we also began to put selected totals into the table. We didn't like to total these tables because there were so many holes in them that we felt that if we put totals at the bottom, people would assume that there was 100 percent data coming in from everybody, so we didn't want to do that. But we were called by the person who was in charge of the HRSA grant at the time and were told that we really needed some data on totals. So we put in selected totals.

We selected on the basis of completion of the data. So if the data were essentially complete in the table, we would put a total. If it wasn't, we'd put a note saying it is not complete because so many states didn't respond that we don't want you to get the wrong impression.

So we did that through the years, up through 1995. Now, these reports ran about three years behind because by the time the programs got the data completed with their diagnosis and the time the national data on birth statistics would come out, it is usually a couple of years behind. We kept trying to get that closer and closer to reality, but we never were able to succeed actually.

1999, this transitioned to become a function of the National Newborn Screening and Genetics Resource Center. So we continued to put these data reports together in 1996, 1997, 1998, 1999, and actually 2000, which I didn't have a picture of. Those we all have in hard copy, and they are available on the net. So people have been going to the net, downloading these data over the years, and finding it very helpful.

Even though in some cases it is not as complete as we would like it to be, it is as complete as the states would provide it to us, and it is as valid as the states would validate it for us. So whenever we would see things that were obviously in error, we would telephone the state and try to get it corrected.

Even with all that, there are still some places where there are obvious errors. But when you call the state, they won't change it, for whatever reason. Usually it is because the person who did the data is no longer there, and they're not comfortable with changing it.

So this is the sort of thing you will find. The 1998 report is what I have given you, just to show you the 10-year difference. It is basically the same information up through 2000 in these kind of reports.

If you look at Chapter 1, you will find live birth statistics. These statistics come from the National Center for Health Statistics when they get their data finalized. It is all dated by occurrence of births, which you have to actually ask them specifically for, because that is not the normal data that they report out. They normally report out births by residence. State laws have to do with where the birth occurred, and not where the baby was residing.

They also have data on Asian births subdivided into five groups, and so we just copied that over. Hispanic births are divided into six groups, which we also copied over. These are always available

on our data set so that you can go look at it. If you need a denominator to do your calculations, this is the denominator that we use.

If you ask the states how many live births, you get all sorts of answers. So we decided we would take NCHS, because that's the official data coming from the state.

Chapter 2 is an overview of the programs which has summary tables of which disorders were screened in the state that year, how many laboratories were doing screening within the state, a listing of the components of follow-up, the basic components, summary of the ages at the time of the initial testing, summaries of the fees charged and the components included in the fee, and criteria for second screening. That is whether it is mandated or whether it is required if it was taken before 24 and before 48 hours, and then miscellaneous information about specimens. How many were received, how many were unacceptable, how long they were stored, is there a policy on storage or not, is there computer information in your program, and what is the form of it, what is the type of education provided, and so on. So just basic information about the program.

The next chapters, Chapters 3 through 13 would be summary data on all of the disorders, where basically each disorder has a chapter. That would include program definitions for the conditions.

Now, this is an issue because every program, as was pointed out yesterday, seems to define things differently. The idea over the years was that we would start off by asking the programs for their data. As I mentioned, for the first three years they wouldn't report data if they didn't define it the way the report defined it.

We changed that to say give us your definition, and then give us your data, and we'll have a table of definitions and we hope that you'll look at other program's definitions and eventually the definitions will coalesce into one definition. That really did not happen. States maintained their individuality like you wouldn't believe. So there has always been programs of definition so that you can look back and see how did they actually define these things.

We have a definition of how the laboratory defines not normal. Again, this came up yesterday. Every laboratory tends to define not normal a little bit differently. It more often than not depends on what the follow-up program can handle in terms of follow-up, so lots of the disorders are tighter down to a certain amount of follow-up. So for instance, for hypothyroidism, most programs titer that down to about .5 percent for follow-up, even though they know that .5 percent is likely to miss a case every now and then. They know also that if you start following up 1, 2, 3 percent of all the babies, the physicians will lose confidence in the program. So there is a balance that has to be made there.

We have the initial screening results, the number screened, the number not normal, the number that were lost, the number that were diagnosed, the number of variants that were diagnosed, and the number of those that were significant or not. We also asked for the same information on second screening specimens. All of this is divided out into chapters so that you can use the data however you see fit to use it.

The diagnosed cases are subdivided by race, sex, and also Hispanic ethnicity. We have days from birth to treatment for all the disorders, and we have summation data from the beginning of when they started screening for that particular disorder. So how many PKUs have you seen this year, and how many have you seen since you started, and what's the date you started?

So where are we now? We have just transitioned on January 1st to an online system called the National Newborn Screening Information System, NNSIS. This has been a system in development for about four years. We actually started as soon as we took over the National Newborn Screening Genetics Resource Center. We convened our Newborn Screening Advisory Committee, which was essentially the same advisory committee that had been working under CORN, asked this committee whether or not we

needed to change the data elements, got agreement on the data elements again, and then got agreement on how to move forward with an electronic online system. Then we put that system out for bids and got a bid from a company and moved forward, but it took them three years to develop it.

That has now been developed and is online. We have trained everybody in the states how to use it. Every state has responded to us with the exception of one state. We have all of those people now online to data beginning January 1st. They have also gone back and they are entering data from 2001, '02, '03 and '04. They have already completed 2001, so now they're working on 2002, '03 and '04. But in the meantime, we have told them to start off with 2005 and be up to date, and go back and catch up on '02, '03, and '04 within the first six months of this year.

So this is what you see if you were able to go to the start up page, which right now only the people who are designated by the state as data enterers can do. But eventually this system will allow anybody who wants a report to go in and get a report from the public or whomever. Users of this system can get special reports, but the general public will be able to get general reports about just about anything they want.

There is the typical security features involved in entry of the data. Certain things they can change themselves if they make a mistake, and certain things they can't. We have staff that does that. Let me just kind of show you. There is basically three things you can do with the system. You can get a report, you can enter data, or you can contact us. It is very simple, as Judy said. It is set up so that Judy can use it, so anybody can use this thing. Even I can use it.

So if you choose the reports, then you get the same reports that you got before with our other system in paper, but you get a lot more of them, and you get them up to date. So you get all these kind of lists of reports. You get overview reports, you get live birth statistics, you get the disorders included in the program, you get the laboratories, all the things I just mentioned, and more. You can get individual disorder reports for any of the disorders that are being screened that are having data reported, which is well beyond the list that we had previously. So we are including in this all the mass spec disorders as we add them.

So if you did one of these reports for a condition, then you'd go to this menu and get all these different kinds of reports for an individual condition. So you could get a listing of all the states and all the information from that state on the laboratory testing results, the timing, all these different things. I'll show you some examples in a minute.

You get also definition tables. So we still are in this situation of definitions, because we still have not national agreement on what the definitions for the disorders are, even PKU. We had a special group come together that were experts in PKU, and we said let's define PKU. This is a simple one. What is the definition of PKU? Well, Harvey Levy was sitting there, and he said, everybody knows the definition of PKU. You've got to have a value greater than 20. Half the room said no, it is 10 in our state, or it is 8 in our state. So no agreement even on PKU.

Again, we have certain things that are divided up by ethnicity and sex. We have maintained that over the years because even though some states don't collect that data, from the states that do, you can make some pretty good assessments of what is going on in the country.

This would be a typical report, just a general report on births if you wanted to look at the birth report. It would go on down through all of the states. So you could print this off. So here is the first 10 or 16 states, all the data that came from NCHS. There is a report here. So anytime you go into one of these reports, it is going to ask you for the year that you want the data from.

Likewise, you get this overview report if you go to an individual state. So, for instance, if you wanted to know what Texas was doing in 2001, then here across the top are live birth statistics, and then

down the table it tells you what disorder, when it started, how many cases they had since the beginning, and how many cases they had in that year. So there are some really nice reports that you can get out of this system.

So if you were to run now a copy of every report possible on this system, it is something like 1,000 pages. So it is a huge book if you wanted to put it together as a book.

Here is another one. Age of the baby at the time they were screened. Again, not everybody collects this data. But those that do, you get some interesting data. It tells you the number that are screened in the first 12 hours, the first 24 hours, day one, day two, day three, and so on. We collected this data already, it is just that we have it in a different format now, and much more up to date.

General information about the program, we still have that available. Again, it would tell you the number of specimens that a state received, the number that were unacceptable, the percent that were unacceptable, the length of time to keep the specimens, and so on.

So there is plenty of those reports. I'm not going to go through them all. Just to give you sort of a flavor for it. I'm also not going to give you the website right now, because we're having the states go through and bring up to date their 2002, '03 and '04 data. Right now what we have done is we've just brought forward 2001 and populated 2002, '03, and '04 for the information about their program, and we know that's incorrect. We wanted to give the states something they could edit, because if we ask them to put in individual data, we wait forever. So we've asked the states to edit that. We hope they'll edit it in the next couple of weeks.

I've just talked to Donna, my able assistant here, and Donna will be calling states who haven't populated it within the next two weeks. Then we hope to have this website available to the public. Right now it is available to the states for their enterers. It will be available to the public through links through our website. So if you go to our website and you're looking for information, you'll get those links, and you can go to those data pages and do whatever you want to do.

We're also asking the states that whenever they find an error, please let us know so that we can correct it. Also the general public if they find an error, please let us know so that we can contact the state and validate it that way.

So what are some of the challenges we've seen? This gets to some of the things that Coleen was talking about yesterday. Things you could be envisioning for subcommittees to work on. Well, right now data reporting, as I mentioned yesterday, except for Title V, is not mandated, or is it a program defined responsibility in most states. They do it because they sort of have to do it to report to their advisory committees, and not all of them see this has a defined responsibility.

The only state right now not participating in our online system is New York. I'm not sure why. They sent us a note the day before the end of the year and said we won't be participating. I sent a note saying what can we do to help, and we have not gotten a response yet. So we hope to clarify that and get New York onboard, because that's a huge chunk of babies, and we need that data.

The national data set has not been mandated. Right now our committee has said where they thought were good data, and the things the states would respond to us with. That has been worked on over the years, and has changed very little, so we think we've got the right data elements. But again, that is not mandated anywhere, it is just a committee consensus.

The data definitions are certainly not consistent from program to program. That was pointed out in the ACMG report as well. There are staff shortages and turnover that relate to data entry issues. So even six months from now if you ask a state about a question in their data, many times they will say I'm sorry, I can't give you the answer to that because the person who did it is no longer with us, and we don't

have good records from that person. I can't tell you how many times we've run into that as a problem. Also they have to understand the data definitions. New people coming in, it takes them awhile to understand those definitions.

Data quality is difficult to validate. It depends on the program. Again, we see things that we think are wrong, and we call the state and ask them to validate it. So most of the time it is pretty good, but there are instances if you look through those reports where you'll have questions.

Maintaining data on border babies or others moving into a jurisdiction are a problem. So if you ask a newborn screening program whether or not they keep up with the data on a baby that was born in their state but lived across the border, you'll get mixed answers. Some do, some don't. If you ask what about a baby who moves into your state, you'll get mixed answers. Some do, some don't. So that has never been really defined well at a national level.

Military births are a problem. Sometimes military births are in one state and the specimens are analyzed in another state. We have been working with the military to develop policies, and the Army has developed a policy. The Navy and Air Force are looking at policies. Those policies basically tell the states the basis to follow whatever the policy is in the state. But policies like that are made to have exceptions. The first thing the programs did in states where they were in one state and being screened in another, they asked for an exception to the policy, and they were granted exception to the policy. So it is difficult to track sometimes military births.

Then we are working on automated downloads. So once you have got a system online, why not automate the download from the state. There are at least two companies who now are selling software to their customers that would allow them to download the case-specific data nightly, for instance, if they wanted to, or the laboratory monthly, weekly, or however they want to do it.

So we're hoping that eventually this will be a pretty automated process where the person in the state won't have to worry about anything except pushing a button and everything will take care of itself. So the idea is whenever a case is diagnosed, then when the person is putting that diagnosis into their records, they just go to our system and fill out the 15 seconds or 20 seconds of data that we ask for so that we have the case in our records. Then that's the case data that you'd be looking at.

It is general data. It does not relate to names. It does not necessarily relate to date of birth, although we ask for date of birth. We also have the option to give us number of days from birth until treatment, or birth until diagnosis. Some states saw date of birth as a HIPAA issue. It assigns a case number so that the state can look back and edit things if they want to. That is the only way there is linkages to the data. That's pretty much our overview, so I'll answer questions.

DR. HOWELL: Thank you very much, Brad.

Are there questions? Lots of questions.

Denise?

DR. DOUGHERTY: Hi. I'm flipping in this book here to the page on program follow-up, and I looked at sickle cell as an example.

You've got a couple of columns, and I'm wondering if these are redefined or have different criteria, or what the criteria are in the new reporting system. You have a yes or no for a program that has an aggressive follow-up program. Program has extra staff, and then you have details on the sickle cell follow-up performed for carriers.

DR. THERRELL: Yes.

DR. DOUGHERTY: I'm just wondering what the definition of "aggressive" is.

DR. HOWELL: What page are you on, Denise?

DR. DOUGHERTY: Oh, I'm sorry. Page 177 in the 1998 book.

DR. HOWELL: Thank you.

DR. THERRELL: I'm consulting with Donna here because she has really worked on this project closely. She says that we have the exact same table in the new data. The definitions are at this point pretty much left to the states to define, although we are trying to drive some of the definitions about case diagnosis. We are not really at this point driving the definitions of follow-up and those sorts of things.

DR. DOUGHERTY: That explains why Alabama says it is aggressive and it has a community-based organization, and Oklahoma says it is not aggressive, or not Oklahoma, but South Carolina.

DR. THERRELL: Right.

DR. DOUGHERTY: It says it is not aggressive, but it has a sickle cell community-based organization.

DR. THERRELL: We are putting exactly what the state gave us right now. Now hopefully, and I think this is something that I think we could work on at a national level, is to define these things better.

The committees that have worked on this over the years felt like at this point it wasn't their prerogative to do that. If they did it, the states would start not answering the questions again, which we didn't want.

DR. DOUGHERTY: Well, I have to say, overall this is a great system.

DR. THERRELL: Thanks.

DR. DOUGHERTY: It seems like the IT-based reporting is a good addition.

DR. HOWELL: A huge amount of information.

Joseph?

DR. TELFAIR: Yes, thanks, Brad. That was a good update.

I think in terms of the issue related to follow-up, if I can make a suggestion to you and maybe to your committee. One of the things that may make it more palatable when you get back and talk to states is just developing a simple criteria for defining what is done.

You can then set some level at which to do that. I understand that there would be confusion that some states, there are about 60 plus programs in the country, for example, for sickle cell disease that are

community-based programs. But not all of them work well with states, and not all of them work well with other things.

DR. THERRELL: Right.

DR. TELFAIR: So it would seem to me that maybe developing a criteria for the level of work they do in relationship to the specific task may be something that is a little bit more palatable to the states. They can tell you whether or not they work closely or not closely, and let them define it. But then you can use that as a criteria to do that. That would actually make it pretty benign from their end, but also give you a good metric to work with.

DR. THERRELL: No, I think that's a good suggestion, although I would hope that there are other groups than us that would maybe do that. These are huge issues, because a lot of states actually contract out the follow-up. That is defined differently from state to state. So there are many, many issues here, and I think it needs to be a fairly large consensus group that does that kind of thing. Particularly with respect to follow-up. I think the idea is good.

Coleen?

DR. BOYLE: Thanks for the overview of your new system. It looks real exciting.

In terms of your last slide with the challenges, what do you think it would take to get states to report in a more timely fashion, and report this data?

DR. THERRELL: Well, we hope we've got the solution to that. Now that it is online and it takes less than a minute actually to put this data in on a case, we are hoping that as people start using it, they will find it so simple and so easy that they will want to do that, rather than keep their own internal systems. If you go to the states, what you find is a lot of them, I mean, more do this than not, have their own little bookkeeping system on their computer. Nobody else in that whole department knows how that system works except that one person. If that person leaves, that data goes away.

So we are hoping that when they see how simple this is, that they will start putting the data in here and then go into our system and pull off these reports. These reports are more comprehensive than anything they've got most of the time. So when they go to their advisory committee meeting, they'll go in with their six-month report that they've pulled off of our data.

So we're hoping that by them giving us less than a minute of work on a case, we're able to give them back stuff that they'll use. Now, we have a system set up within the computer that alerts us. Right now we can set the time, and we've set it at 30 days. So if in a 30-day period somebody has not put any data in, it will send them an email and it will say you haven't put any data in for the following conditions, because in some states one person may do some conditions, and one person may do a different condition. If you haven't put it in in 30 days, it will say we've had no data from you in 30 days. Does that mean there has been zero cases, because zero is a real number, and we want to bring it up to date. So if it does, then please go to this place, push the button, and it will put a zero in for you for all of your disorders.

If they don't respond to that, then my policeman gives them a call after a certain number of days and asks them if can she do it for them. And so we are trying 30 days. Thirty days may be too tight, but we thought that was reasonable. It may be we have to back off to 60 if they start complaining about it. But so far we had this beta tested in a number of states. We didn't beta test this particular part very much, but it seemed to work. Maybe Dr. Becker can tell us how his laboratory and his follow-up would respond to this. But we're hoping this is the kind of response that will bring this stuff up to date.

DR. BOYLE: Just a comment for the committee. I mean, we do birth defect surveillance system reporting, and you'll hear a little bit about that from the next speaker. In order to get states to report, we have to pay them to do that. So it is an ongoing challenge to get timely information. I mean, it is a wonderful system as you've outlined it.

DR. THERRELL: If it works like I've outlined it.

DR. BOYLE: Exactly.

DR. THERRELL: I mean, we're hoping that it will pay for itself because they will get these reports that they want, and they'll see that as a real plus for them. It remains to be seen at this point.

DR. HOWELL: Piero?

DR. RINALDO: Brad, I see this presentation as having a dual purpose. One is to educate the committee about I think an extremely important initiative. But I also think that it could also be an opportunity for you telling us what we can do to help. So I think this has been asked by others, but can you be specific? Assume it is Christmas. What would be your list?

DR. THERRELL: Well, you know, to help out with the data, if there were a national mandate of some sort that states had to report data at certain times, in certain ways. That would be the ultimate thing that I would want to see.

Now, in order to get there, you have to have agreement on definitions, you have to have agreement on the need for the data, you know, what are the appropriate fields and that sort of thing. So that is where I see a committee sitting down.

I don't think we have to worry too much about the fields, because that has been debated for 10 or 15 years now, and everybody is pretty much agreed that the data we're collecting are the data that they'd like to see collected and the data they would report to us, with a few exceptions. Some people say, I don't have the race data, and I don't have the sex data. But most people have that, so that has been pretty well worked out.

What hasn't been worked out is why do I have to do this, and why is it to you, and why is it just definition? Those kind of things. So I'd like to see that done. Title V does some of that, and I'm not sure there is any particular federal agency that can mandate this. But it would be nice if somehow it were mandated.

DR. RINALDO: And, you know, I can see certainly this being something for one of the subcommittees that we're going to create to really address, because I agree with you. It certainly would be a major incentive if state programs were aware that at the end of the year, that report that happens to match, the ones generated by this, are required.

DR. THERRELL: Yes, I'd like to see it more than just required at the end of the year. I mean, yes, a summation required at the end of the year, but I'd like to see it sort of required within 72 hours of you having a diagnosed case, it goes into a database somewhere so that we can track up-to-date information.

DR. RINALDO: But I do want to warn you that I actually think that 30 days is probably not in your best interest. If you look at programs, on average programs depend on the number of births they handle, but they can handle a few hundred to thousands a day. So that means that defining of collectively all of the condition screening, if not a daily, is certainly a weekly event.

So if you wait three weeks, four weeks, an entire month, you can be certain that it is already old data that hasn't been entered. So I'm wondering if you might want to consider being a little more aggressive.

DR. THERRELL: Well, we've debated that back and forth, and maybe we do, but we're going to start off with 30 days and see what happens. We think that most people are going to report them as they diagnose the cases, because most people put those into a notebook somewhere and put them into a computer system. This only takes a minute.

That's the problem. We're having to convince people it really does only take a minute or so. So when they use it, which they are now using it, every state with the exception of one has used it and has agreed that it does take less than a minute to put a case in, and they like it.

DR. HOWELL: Bill? A comment from Bill, and then from Steve.

DR. BECKER: Thanks, Brad.

We have experience with it now. We hope that, as Brad indicated, that it is easy to use, and as much as possible for us, we're going to enter the data in real time, which is certainly the hope. Time will tell if the 30-minute window is the appropriate window or not. So it is sort of a pilot project going forward, which is fine.

I think it may be a little slower in the

start-up period for reasons that Brad also mentioned. Not only are they doing 2005 going forward, but we are also playing catch up on a couple of years, too. So it is going to take a little while before we know the answer to that, but I've seen the screens. I'm not a user, which is a good thing, but it seems intuitively friendly enough.

My customer service area, my follow-up people, I believe that it should be pretty helpful to us. I would differ with Brad on the concept that it would ever replace our internal database. Although it might replace portions of it, but I think for some sense of safety, at least for awhile, obviously the concept of duplication will drive us to reconsider that at some point. It is a potential.

Brad, a couple of questions. I think this is a huge effort, and I certainly would support committee activities or subcommittee activities around this issue. To me, information management is absolutely critical for newborn screening programs.

I see some challenges for the states that might need some resources. Some states have a separation of laboratory services and follow-up services, such that the databases are not the same. They are not even shared, and they might not even be talking to each other. So communication with this information system could be a challenge that the states need some resources to address, which also might delay them in their reporting practices as well, depending on how they are structured.

I also see some challenges while most laboratories will probably be able to solve the issue of electronic reporting through the various software packages that you alluded to, I'm wondering about the challenge of private laboratory reporting, and getting data from there, whether that would be a pass-through through the state, or whether some direct form of communication would be possible.

Then finally, which to me is the single most important aspect of this project, is just like we're concerned and focused on the issue of laboratory testing quality, the issue of data quality is exceedingly important. The validity of the data almost brings to mind a rigorous process. Again, I am harkening back to the cancer registry and the types of personnel and the education and training of the personnel for the data entry that is needed to accomplish those tasks.

There is a lot of databases in health departments, there is a lot of electronic communication going on. We're getting lead data reported to us from private labs. We have solved a way to do that. It took a

little while to do, so the challenges are there. As some of you might know, state health departments are also working on electronic reporting for infectious diseases, as well as any of the other reportable diseases that the state has to deal with. So newborn screening is certainly one of those challenges. It is an important one, as I think we'd all agree.

DR. THERRELL: Yes. A couple of comments. We do have separate menus for laboratory and follow-up for certain things, and we have different people allowed to do different things with those data, so we've addressed some of that.

The laboratory data that we're asking for is pretty general data. It is available daily, but most laboratories don't like to report it daily. With automated software, maybe they would. We're hoping that.

The other thing is we have talked to private laboratories. We have built into the system a way for the private laboratories to download data for the state, so they have multiple state entries that they can go in. For instance, if Pediatrix were entering data for their customers, they have the District of Columbia, Pennsylvania, Mississippi, and Nebraska, they could go to the menu for Nebraska and download the data for Nebraska, go to the menu for Mississippi, download the data for Mississippi. In fact they have done that to us on paper in the past.

Generally it was a pass through the state, but in certain states, they preferred for us to get the data directly from the private laboratory, and in certain states they didn't have the data, and that was the only place we could get it. The private laboratories have been very good about cooperating with us on data, so we have a mechanism for that. We are working on that. We haven't yet got that in place, but we don't think that is going to be a big issue.

DR. HOWELL: Dr. Edwards?

DR. EDWARDS: They're still patients, not customers.

DR. THERRELL: Sorry.

(Laughter.)

DR. EDWARDS: I'm wondering if in your vision, and I think this is a great start, and I realize that this is group data, anonymous data, and all of that. But with the recommendations that we are coming up with in this group, we are going to experience a new problem, a problem that we've had, but a problem that is going to be more serious. That is for the individual practicing doctor with the increasingly mobile society that we have, do you envision this in any way in your wildest dreams evolving to a kind of system where doctors can turn for patient data?

Because so often state data is not enough. People move across state borders and doctors need information about the patients. It is going to be more and more so with more conditions becoming available. Have you had any vision about this?

DR. THERRELL: Well, in my wildest dreams, it would be possible to do that. But I think in reality given all the privacy issues, it is going to be a long time before we're able to do that in a national database. I mean, this was tried one time before.

Susan Meese had a database that kept up with the metabolic patients, just so that metabolic docs could follow the PKUs and MSUDs, and that was a real problem. It now doesn't exist anymore, I don't think. So I think the states will be able to maintain that. This system was set up to get the gross data from the states, or to get specific data that didn't link to the patient that could be used in a gross data sense. So I think it will continue to do that. It has the possibility to do what you say, but I think we've got

to really address these privacy issues that Dave talked about and Judy talked about yesterday. So that's a long way from now, I think.

DR. HOWELL: Greg?

DR. HAWKINS: Your comment on data on military births kind of piqued my curiosity. It may be a little off subject, but what about families that are in other countries for the military? What happens to the children? Are children tested there? And what about U.S. citizens that are in-country? Do they have access to tests? And is that information still reported to you guys? Does it get into databases and stuff like that?

DR. THERRELL: It happens in variable ways. In the military, a lot of the different military bases overseas have mechanisms in place to send those back to the states and have a state do those. For instance, Colorado does a lot of those, Oregon does some of those, and Texas does some of those. I don't know whether Ohio does or not, but there are mechanisms in the military to send those back.

Now, U.S. citizens traveling abroad, the ones that I have talked to who have had babies have generally done it with a local program, which didn't always cover all the disorders they wanted. If they knew better, they would even have specimens sent back to the states. But again, that varies from individual to individual. It is not widely known how that happens.

I can tell you that there are a lot of programs around the world, a lot of good programs around the world. Even in the military procedure that the Army set up, it says that they can use a local laboratory. For instance, in Korea, they are using a local laboratory which participates in CDC's proficiency testing program. So there is a worldwide network of newborn screeners around.

DR. HOWELL: Joseph?

DR. TELFAIR: Yes. Brad, just on the question of utilization, this is a two-level question. The first level is at the aggregate level. In your advisory committee and then what you are working on, have you all begun a conversation about the political and ethical use of the information? What I mean is if the data is aggregated and those who are interested in developing policy are developing services at the national level and that sort of thing. Then also ethical in terms of who also has access to this database besides just the states. Have you all begun conversations about that? That's my first level question.

DR. THERRELL: Go ahead.

DR. TELFAIR: You can go ahead and answer that.

DR. THERRELL: The first part of that is no, we haven't really worried about who and how they would use the data. We rather put the data together on the basis of this is what the states felt would be useful to them and to the public, and this is the data that they felt comfortable that they could report.

We have had discussions about who would have access. So we have this set up to make this available to the public in a general format so that all those little reports that I showed you, you don't have to have a password to get to that reports menu. You could go in there and generate any reports you wanted over a time period you wanted, and theoretically use it however you wanted, although most of the time it has been used to prove a point or to move a program forward as opposed to moving a program backwards.

DR. TELFAIR: Well, I bring it up because a lot of times when you work on these large data sets, the questions always come up about appropriate and inappropriate use of information and by whom. So that's why I was asking.

The other piece of it is you sort of answered the question on how is the information utilized. I was wondering if in the fields that you're thinking about or that you have, would you begin to look at how this information is being utilized by the states for decision-making, or if not by the states, then by others who are trying to make decisions.

DR. THERRELL: Yes, I think it would be good to do that in a set way. We do it now anecdotally. We hear that a state used it this way, or we hear that a legislator used it this way. Believe me, they have been used by legislators. We get calls a lot from legislators wanting to know why is it that this state does it this way and our state does it this way, and can you help me understand what is going on here because we've got a bill pending and I want to be able to answer those questions.

Likewise, the reason we did this 10-year data which we're trying to get together to publish is to show you trends across the country. If you look at that data, what you'll find for instance with hypothyroidism is that the incidence that used to be reported to be about 1 in 3,000 over the years has gotten higher and higher and higher, and it is now about 1 in 1,700.

Now, if you start looking at why that is, it turns out that in the data collection that we've been doing, over the years we've gotten closer and closer to real time. So we have a lot of transient cases included in the data that don't get taken out.

Not only that, it appears that the practice of medicine has changed a little bit and that the pediatricians don't go to the trouble to go through the in-depth analysis that they used to go through. They just go ahead and put these babies on treatment if they get the results back and then worry about it at about two years, take them off, challenge them, and see if it was transient hypothyroidism.

Those numbers are all getting reported back to the state, and the state is not cleaning those up because we're getting the data too quick now. If we get the data at two years or three years, we can clean that up. We've got to look at that. That is an issue that we're going to have to look at for hypothyroidism. It doesn't seem to be a problem for other diseases, but there are lots of ways that you can use the data, and we don't want to restrict anybody right now.

DR. HOWELL: Denise, and then Piero.

DR. DOUGHERTY: Yes, this is related actually to uses of the data, it is a question. Yesterday about the secondary screening we discussed, and I see that a number of states do require it, I'm wondering if these data could be used to look at that question of how many are confirmed with secondary screening. You would need the cut off values and so forth, and I'm not sure you collect that.

DR. THERRELL: No, we actually do collect what the laboratory uses as a definition of not normal, which is the cutoff value. Some people have used the data exactly like you have said. They have used it to go back and look at second tests in the state, and they have used that data to convince their state to go to a second test or not. So it has been used that way.

Phil Farrell wrote an article a couple of years ago where he actually took these data and looked at them and made some suppositions which I considered not to be too good because he related it back to the tests themselves. I mean, you have to understand the data and how it is collected. He related it to the tests themselves, which is not correct. There is a process involved, and the state may have decided on a cutoff not because of how the test necessarily was running, but more what the follow-up group could handle.

As I said, thyroid was titered down from about .5 percent, or 1 percent in some states, and it wasn't looked at in terms of what is the cutoff value that we're using, let's talk about what is the percent

our follow-up group can use. So all of that plays into those kind of issues when you start looking at those data.

DR. DOUGHERTY: Is that part of your mandate, to analyze the data in that way?

DR. THERRELL: No, not right now.

DR. RINALDO: Well, I want to follow on the same topic. Sometimes you have to pause and reflect. You might be collecting an enormous amount of data, but then they just sit there. I'm wondering, because you certainly have raised, and a number of times we have talked about the issue of the quality of the data.

I really think there should be a way to identify areas of potential improvement. Just open it randomly at page 100. When you see that for initial screening results for homocystinuria, you look here. Indiana and Massachusetts have reported a comparable number of screens. They both had no true positives. But once they reported double the number of abnormals.

So this is the kind of data where there should be a way to say, you know, there are just too many. So there is a root, an analytical root, behind that. I also see it as an opportunity. I think it is a very legitimate argument when we try to change something, or to do something differently, we need resources.

I really think there is an obligation to really revisit resources that are misused. So it is quality and performance. Clearly I see this is a process. So I think it just makes sense to start from the early stages. I often go back to the testing.

But we must address the issue. One of my favorite activities is to question the wisdom of the screening. That really leads to kind of an acceptance of performance that is really probably not adequate. Data like this, just opening it randomly, are really very obvious. So I hope that this data once collected must lead to an assessment of performance, and not as a judgment or a trial, but very much as an identification of areas of improvement.

This improvement can result in cost savings, meaning there will be resources available for other activities. Do you agree?

DR. THERRELL: I agree. There's no national mandate to do that now. Internally within the states, if you are smart, you do that, and people do it all the time. When I was with Texas, I did it all the time, too. If I saw something California was doing that looked better, I'd call California and ask them, why is it your data looks better than my data? What are you doing different? We would go through it. Or New York, or whoever.

I have been to states where they actually have set up their internal quality assurance based on these data, looking at their data over the years, and looking at their data relative to other states. Now, you can't necessarily look at these data and say that means bad analytical technique, because without talking to the state and finding out how they decided to use that cutoff, you don't know for sure if they decided on a policy basis that they wanted to see more positives. So everybody's definition of how you screen and what a screen positive is is a little bit different.

DR. RINALDO: I agree, but remember, in September we heard a presentation by Cunningham and by Ken Pass. To me, it was really interesting to hear that in one state, the follow-up on average cost about $1,000, and in the other state they were down $259. Nevertheless, we are talking about in just really our example of 50 cases that it could be $200 or it could be $1,000. But when you put this on a national scale, it is a huge amount of money.

DR. THERRELL: I don't disagree.

DR. RINALDO: And so it cannot be continued. There are things that say well, it is okay, screening, so it is okay to have a very high number of false positives. I don't really think it's acceptable.

DR. THERRELL: Right now, that's a state by state decision.

DR. HOWELL: Coleen?

DR. BOYLE: Just a quick point. You've talked about the concept of mandated federal reporting. In all the reporting that we do at CDC, there are very few conditions that are mandated at the federal level, most of them are state mandated. Then we work with states, and I think that is what you're trying to do.

I'm not sure we would want to recommend as a committee that there be mandated federal reporting here. I think that the reporting and the responsibility for follow-up really resides at the state level, and we want to help facilitate that. I think all the issues that you and Piero were just talking about are really very important and very critical. That is really what the data is for is to make the program work more efficiently and appropriately, but we want that to be done at the state level.

I see you as being the facilitator of that, helping develop the standards, you know, maybe centralizing some of the reporting so that we can compare across states and help with those case definitions, help with program monitoring. I can't imagine mandating the reporting on a federal level.

DR. HOWELL: Bill?

DR. BECKER: Yes, I want to follow up on that. There is a process in the states. I agree with Coleen, but there is a model that we could consider at least patterning a national reporting process for newborn screening after.

That is you pick up the MMWR every week and you go to the back tables, and it is the national notifiable infectious diseases. These are infectious diseases that have been agreed upon by a number of the partner organizations. CDC, ASTHO probably inputs a little bit into it, but mostly the Council of State and Territorial Epidemiologists that produce these lists of infectious diseases that all states should be reporting on.

So we might consider now is there a national mandate or a federal requirement that states do that? The answer is no, but I think there is a pretty good acknowledgment at the state level by working with the state partners that they get reasonably good reporting response from that.

So a national notification or national database of inherited or heritable disorders or newborn screening disorders or whatever the name we want to call it, could be patterned after the infectious disease model that has worked so well for many years.

DR. THERRELL: Well, what I was talking about was sort of tied to Title V. That was a model that is sort of already there, already certain things that are required to be reported under Title V, because most programs get some Title V money in the mix within the state. Title V is MCHB's, the block grant to the states for maternal and child health services.

Within that grant are some funds that are available for newborn screening. Most programs use some of those funds, so there is sort of a way to do that as a requirement. It just needs to be expanded in my mind, and you'd have it. If you ask states to voluntarily report it, I think that some will and some won't.

DR. HOWELL: Brad, let me thank you for an excellent report. Obviously acquiring and getting all this data widely available is going to generate a tremendous amount of interest and activities for improvements in research and the whole nine yards. Thank you very much.

We had better move on to New Jersey. We are going to hear about the birth defect surveillance system from the New Jersey Department of Health from Leslie

Beres-Sochka.

MS. BERES-SOCHKA: Hi. I'm Leslie

Beres-Sochka. I'm from the New Jersey State Department of Health and Senior Services. I'm going to present some data on our screening surveillance and service system that we've developed in New Jersey. It is slightly different than what Brad has been talking about.

First off, I'd like to present some national data compliments of the Centers for Disease Control and Prevention. About one-third of all live births have a birth defect, which amount to 30 percent of pediatric admissions, at a substantial cost to the economy. There was an MMWR article in 1995 that listed the 17 most significant birth defects, and they estimated the costs of caring for these children to be $6 billion annually.

Some of the causes are preventable, so it shows that if you work on prevention, maybe we can lower some of the economic costs for these children. It is the number one cause of infant mortality in the United States, far exceeding all the other common causes of infant death.

The history of birth defect surveillance is in the 1960s there was a lot of international interest due to the thalidomide incident. Metropolitan Atlanta started their program in 1968, and by 1974 there were three additional programs, one of which was in New Jersey. Now we are up to 38 operational programs. I think it is 37 states and one territory, and there are nine in the planning stages. Here is a map showing the distribution of how all the states fit in. Five have no program currently, which is something that I believe CDC is working on.

This past year in the fall, CDC surveyed the states on their birth defects ability to refer children to the service system. Of the states that replied, 26 had a system in place already, 17 were planning, and 3 had no plan for linking the birth defect surveillance to the service system.

Of the ones who were linking to the service system, there was kind of a variety of what they were doing as far as using their data. Some states referred everybody out, which is what we do in New Jersey, and some states just had some specific things like neural tube defects or cleft palate, they would refer those to services, but they didn't refer the bulk of the other services. You can see from this list here how very different it is from state to state. I have some of that data if anyone wants to see it later.

I'll change hats now and go back to my state. We are in the Health Department, the Division of Family Health Services, and we have our Special Child Health, WIC, Maternal and Child Health, and then an MCH Epidemiology Unit. The Birth Defects Registry is my group. We're under our Special Child Health Services and Early Identification and Monitoring. My group, Early Identification and Monitoring, does the Birth Defect Surveillance System and the Newborn Hearing Screening Program.

We used to also do newborn screening and genetic services, but with the big expansion of universal hearing and the big expansion we had in New Jersey of the genetic and metabolic screening, we split them into two groups, so that's why they are separate here.

We have our family centered care, which is children with special health care case management units, which I'll describe a little bit later in the talk, and then we're also an early intervention system. So it is an interesting opportunity in New Jersey where you have four very important components of screening, surveillance and services, all tied into one program. It really enables us as the registry to serve as the linchpin from diagnosis to services. We can do it because we all report to the same agency. So a lot of the issues that other states are faced with, we've avoided by our structure.

Birth defects history in New Jersey, we have a very long history. In the late 1800s we began looking at children with orthopaedic conditions. So by the end of the 1920s, we actually had a requirement to report crippled children. I know that's a very politically unacceptable term today, but that is what they were called, the Crippled Children's Program.

It was mainly for children who had orthopaedic conditions. They had some very forward thinking folks in the 1920s in New Jersey, and they felt that you needed to count them, but you also needed to make sure that they had services, so they set up a whole slew of services for the children. But again, it was just orthopaedic conditions.

Then over the intervening decades, all the different federal initiatives with Medicare, handicapped children, SSI programs, had an impact on the children that we could serve.

Due to environmental concerns and then other concerns with wanting to do more than just orthopaedic children, in 1983 we decided to implement a law with a population-based surveillance system. So we had a law that requires now reporting of children diagnosed through age one of any birth defect condition. It is pretty much the entire range of birth defects. Included in the law, it is very interesting, is the metabolic conditions and hearing information.

We adopted rules in 1985. The purpose of the law was to again, establish a registry so that you could do surveillance and epidemiological studies. But most importantly for us is to plan for and provide services, which is actually the text of the law. So it does enable us to use the registry as the populating base to then refer children onto services.

Under the wording of the law, the Commissioner of Health can give access to records to other agencies. So we have this letter that is getting yellower and yellower dated 1983 that authorizes the registry by the current commissioner at that time, Dr. Goldstein, to share that data with our Case Management Program, which is our Children with Special Health Care Needs Program.

Our rules require reporting from a variety of different sources. Hospitals, physicians, basically we cover anybody who is going to see that child is required by our rules to report to us. Hospital licensing standards have the birth defects reporting and also hearing screening reporting as a check box. So if they don't report to us and it has become an issue, we'll go to hospital licensing to make sure that the hospital addresses their need to report. It is a nice stick to have in the back pocket. For birth defects, informed consent was not required.

We have two components to the registry. We have the birth defects which are the mandated conditions, but we also track and monitor children with special health care needs. That is voluntary, and it requires parental consent. We annually have about 114,000 live births. It varies from year to year, but that's about the average. We get 8,000 new registrations or more every year to the registry. About 65 to 70 percent are the mandatory birth defects. The rest of them are the voluntary special needs conditions. Again, we reside in our Special Child Title V Program.

The registry itself is funded by the Maternal Child Health Block Grant, so I thank those of you sitting here. We have a CDC cooperative agreement, and I thank CDC. We're developing a new database, an electronic reporting system with CDC funding.

We do have a quality assurance plan in place for our registry. I have a nursing staff that goes out every year to every one of our 64 birthing facilities, and they do an audit of three months worth of births, and we vary the months from year to year so that they don't know ahead of time and can't guess like okay, it is June, July and August, we know they're going to look at that. So we keep them on their toes that way.

We do a three-month review. At the end of the review, we have a meeting where all of the hospital administration and the pediatric disciplines that touch that child are required to be in attendance. We explain to them how the preliminary results look. Once we get back to the office, we clarify any misunderstandings or things that we thought weren't reported that were, or vice versa. We do provide a written report to the facility.

One of our new initiatives is we are working on, I don't want to say a report card, because it won't be a report card, but it will be a more formalized report than what we're currently doing.

We linked to other databases as part of our QA. We linked to the birth/death files, and we review all infant and all children's death manually. We get a copy of their death certificate through an agreement with our Vital Statistics Bureau, and we review all the deaths for children under three to make sure that we didn't miss anybody that died, or the hospitals didn't miss reporting somebody.

We do have reporting from other health care programs. Our newborn screening programs, both hearing and biochemical, report directly to us anybody who is a positive. Our data from our QA with the hospitals indicate 80 to 90 percent of children are appropriately registered. If we find someone who falls below 80 percent, we do a year long audit. If it is consistent from year to year, that's when we go to hospital licensing.

We have an EBC in New Jersey, the electronic birth certificate, for those of you who don't know that terminology. It is a comprehensive database in New Jersey. It is not just the electronic birth certificate. It is not just the birth certificate that is required reporting. It is the entire perinatal, maternal, and child stay in the hospital records. So we have this wonderful resource of all this data that is reported to us through the electronic system.

We get the data through an agreement with vital statistics. We are allowed to use it to amend our data and to do research and whatnot with it. However, when we share our birth defects data, we don't share the electronic birth certificate data. That would be a violation of confidentiality and HIPAA.

I'm going to switch hats now and talk about the other component of the program that feeds into the registry, the biochemical screening program. We were screening for PKU legally effective in 1964. Over the next decade, we added hypothyroidism and galactosemia. In 1990, we added all the hemoglobinopathies. We have a law that requires newborns to be screened unless they have a religious objection. We don't get informed consent because of the rule and the law.

With the advent of the new technology, a lot of public concern of issues concerning the availability of screening for more and more disorders, we convened an advisory panel in April of 2000. The panel consisted of a wide variety of medical professionals, parents, and experts in the field. I believe some of you in this room are on the panel. We reviewed approximately 30 disorders at that time. In December of 2000, the panel prepared a report that was formally presented to both the Commissioner of Health and the Governor of New Jersey.

We decided to expand our screening based on that report. Instead of doing everything all at once because of the follow-up complexity, we rolled it out over the three years. So in 2001 we added four, in 2002 we added six more, and then in 2003 we added that whole group of six organic acidemias.

In 2005, we are going to be reconvening our panel starting in March. We are going to be looking at the non-mandated conditions, including this whole long list here, which I won't bore you with the names. I'm afraid I won't be able to say them. But we'll be looking at each one of those with this advisory panel to see what they think is feasible for us to add to our screening and follow-up program.

Our screening in New Jersey, we have the Inborn Errors of Metabolism Laboratory which receives a specimen, does the testing, does the QA on the testing, and our newborn screening and genetics follow-up program. Again, the lab does all the testing and quality issues. They actually report the results back to the hospital.

We have the follow-up program which is in my program, and we actually do sit adjacent to each other. So it's not like we're in the same division of the Health Department but we never see each other. We actually work very closely with them. We sit right next to them. They make the recommendation to the families and the doctors for retesting. They do the case follow-up to final disposition, and they provide a whole bunch of different information to the parents and the health care providers. Again, this linchpin, the registry. Anybody who is a finally confirmed case, they immediately register with the registry.

It is unfortunately at this point a paper process, but they actually just walk over and hand it to us. We immediately get it into our data system and up to the service stream.

We have access to treatment through our follow-up program. Special Child Health Services provides grant support for the following things. We have the metabolic formulas that will provide metabolic centers, cystic fibrosis centers, and then we do have a list of pediatric consultants and the groups for the different conditions. Then we have a comprehensive list of all the consultants in the state and what their speciality area is, which we will send out to the physicians and the families upon diagnosis.

For fiscal year '04, today we have reported 113,414 tests of which over 6,000 were referred for follow-up. Over 3,000 were abnormal, and in addition, there were 3,000 sickle cell traits. Again, the program will follow those to disposition. We don't have the final data yet for fiscal year '04, but so far we have 271 cases confirmed of all the disorders that we screened for. It will probably be much higher than that once we get all the final information in.

They do quality assurance, they monitor time to treatment, they conduct visits to the hospitals. One of the things we did this year was conferences to educate everybody. The Blood Spot Screening Program partnered with the Hearing Screening Program, and together we partnered with our hospital association in the state to present a one-day seminar on changes in our newborn screening policies and procedures. All the hospitals in the state were required to send a representative. I think there were a few that didn't, but it was very good attendance. One of our panel members, Dr. Howell, was a speaker at that event.

I'm going to switch hats now to a program that I'm responsible for, the newborn hearing screening program. It is required by state law. We had a law starting in 1977, it was a very simple law that just told the Health Department that they had to have a screening program for hearing, and we were free to set it up how we chose. So at that time, the best you could do was the risk-based screening. So it was all paper and pencil risk-based screening.

Over the years with the advent of technology, more and more hospitals were starting to do the electronic hearing screens. We had some difficulty getting it into our rule adoptions. But in 2000, we amended our rules to require the phase in of the electronic screening. The legislator one upped us by passing a whole new law which became effective in January of 2002, which it took a lot of what we had in our rule structure and turned it into law. It mandated universal newborn hearing screening. Again, it is the 1-3-6. You screen by one month, diagnose by three months, and hopefully enrolled in some sort of appropriate intervention by six months.

The law mandates universal screening testing by 30 days. The hospitals have to have a protocol. In New Jersey, we can't tell hospitals exactly how they have to do medical things. We have to tell them that they have to do screening. We could tell them what the standards are or best practices are. So we could tell them that a two stage screening is the best practice, but we can't force them to do that.

So to keep track of what each hospital is doing, they have to provide us with comprehensive protocols and procedures, including their responsibilities for follow-up of the children who screen positive.

Eventually when a child gets diagnosed, any hearing loss, including very mild unilateral hearing losses, are required by that statute to be reported again to our birth defects registry. So again, the birth defects registry is serving as the central depository for data.

We also had insurance coverage provided in the law for this screening. Again, it is only the insurance plan that the state is allowed to regulate. So there is a whole group of insurance plans that are not covered by the law. But we are hoping, and we have seen some of them come around. Even the ones we don't mandate because of peer pressure, they don't want to lose their clients.

The hospitals are responsible for both the screening and the follow-up according to the law, which makes it kind of interesting. When we have 64 birthing facilities in the state, it is quite conceivable we will have 64 different follow-up procedures, so we have been working on that. We are using our universal hearing screening money that we have from HRSA or the HRSA grant. We have two staff who has been charged solely with

follow-up activities. They have been out there with some standards and procedures trying to help the hospitals do similar things.

We are in the process of a new rule adoption now for hearing screening. Hopefully the rules will take effect in March or April. We have spelled out what you must have for follow-up. Again, the registry is central to all of this.

We have an EHDI surveillance system. The EBC, the electronic birth certificate in New Jersey, one of its functions is it does all the risk factors for hearing, and it has the hearing screening information in it. So through an agreement with our Bureau of Vital Statistics, we use the electronic birth certificate to initially populate our hearing tracking system, which we developed through the CDC hearing agreement.

This past year, starting January 1st of this year, we have a whole slew of new variables on hearing screening. Up to this point we just had very simple measures. Did you screen the child, and did they fail. That didn't really give us a lot to go on and to track. So we have now right and left ear-specific data, dates of screening, ear-specific failure or pass rates and so on, including native language for mother, or for primary care provider for the child so that we know as we address their needs, which language would be best served to them.

Again, the EHDI system, one of the things we're working on is linking to our early intervention data system. We had hoped to be online sooner with that linkage, but our early intervention system had some vendor issues. Last year they had to scrap their plan and start from fresh with a new vendor and a new system, which they are currently populating with data. So hopefully this year we'll have that electronic link from the hearing screening registry into the early intervention registry. Now we are doing a manual match of children.

Our hearing screening data, sometime this year, we're not sure when it is going to be rolled out, is going to be part of the database that the immunization registry will have. The immunization registry is now legally mandated in New Jersey to track and monitor the children who get their immunizations.

One of the complaints that we had from the docs is it would be nice if they knew what the children's hearing screening data was. So we worked out an arrangement with the immunization registry. I'm not sure if it is going to be a little pop up box, or if it is going to be just another page that they can look at, that they'll be able to access their children's hearing screening results.

This is just a quick schematic of the database. I'm not going to keep that up there. That's not very exciting. The flow process for hearing screening is the usual. Currently we need parental consent because of the old rules. As soon as our new rules are adopted with the new law, we will no longer need informed consent. But until the new rules are formally adopted, we couldn't eliminate informed consent for the parents.

Moving onto our case management program. Now again, this is another piece of our special child health program services. The purpose of our case management is to assist children through their 21st year to access a family-centered culturally competent coordinated services for children. It is decentralized. We have one in each of our 21 counties. That way, they can reflect the uniqueness of their areas, and they know where the services are as opposed to being state driven where we're sitting in Trenton and don't know anything about the local surroundings and where the best or most efficient places are for the locals to go to.

It serves as the single point of access into a wide variety of services for the children and their families. We fund it jointly from our Maternal and Child Health Block Grant fund, and we have state and county funds that also are involved in funding the local units. In 1993, our case managers began to serve as the single point of entry for early intervention, so we have all these things tied in. The register gets these case managers the data, and then they can appropriately refer the children to either case management medical services, or if they require early intervention, they serve that immediately.

Our registry tries to refer the children out to the case management within 10 days. If it is an urgent health care need and we know the family needs services immediately, we will make a phone call. We have trained nurses on staff, master's level pediatric nurses. They will call the case management unit in the child's county of residence immediately and tell them we have a child that we haven't had a chance to process yet, but they are going to need services. We'll work on getting the letter out to the family, and the case managers will get to work on that immediately. So it is not that every one sits for ten days. The most urgent ones we do make sure get through the system quickly.

The focus of the primary care provider, we make referrals for identified needs. They develop individual service plans appropriate to each child and each family. They also help the families learn to advocate for themselves, which is a very important process.

We have a lot of health care resources that we link the families to through our case management system. We make sure that all the families in our system have access to a medical home, so that we're coordinating all of these other services with the medical home. We have referrals to other agencies in this state. New Jersey FamilyCare is our CHIP program. So if the family doesn't have medical insurance, the case managers are right on top of that, and they make sure that they get enrolled into that. If they are eligible for Medicaid, they work on that, or SSI. It is incredible what we can do for our families.

We help them with some of the prosthetic devices. Special Child Health Services in the state have some money set aside for what we call the appliance plan. We will help them to provide hearing aids and prosthetic devices. It is on a sliding fee scale based on need. We have a small pot of money to provide assistance for asthma and cystic fibrosis medications. We help them with rehabilitation resources, and then again, the advocacy where we link them to our parent-to-parent support group. We have a statewide parent advocacy network, and the subset parent-to-parent, which we work very closely with.

One of the things we're doing with our hearing screening money that HRSA has given us is we funded our parent-to-parent to provide more families who are competent culturally to help be the parent partner for children who are deaf or hard of hearing. We just yesterday hired a Chinese speaking individual, so we're very excited about that, because that's a big population now in New Jersey. We didn't have anyone who could speak that dialect. So now we have a parent trained to work with that population. We also have Spanish, and I think there is a Haitian Creole person, and we're working on a few other of our population groups.

Early intervention system. Again, it is part of this whole process that Special Child Health Services are the point of access into that. The service coordinator will work with the families to develop the IFSP, and to make sure that they receive all the education and all the other services that they need.

In New Jersey, and I don't know how it is for other states, but any hearing loss is a presumptive eligibility. So again, we are tying all of our programs together and making sure that the children who we're dealing with have appropriate services.

This is a flow chart of case management. It is a little complex. The panel, you have it in your folders, so I'm not going to go over it. But if anyone has any questions, I'll happily work on it. But you see there is the registry and case management. Service coordination or case management, depending on if you're eligible for early intervention. Then if you are early intervention, you go on this side, and if you are case management, basically it is the same thing, it is just which group you fit into.

Now, the birth defects registry we use for a lot of different data types of things aside from our service component. We use it for surveillance. The state that identified the Accutane was related to birth defects. We do it for internal reports as well as national, and we participate in a lot of multi-state surveillance reports. We use it for need assessment for our block grant and for our case management units so that they can plan for services.

We had a lot of research projects, which I'll talk about in a minute. Some collaborative projects where we worked with AIDS in our developmental disabilities program, which is in a different department of the state, and then with our folic acid folks, and again, the linkage to services.

These are some of the research projects that we've participated in in the last five years. The Center for Birth Defects Research and Prevention, which we're no longer participating in. The National Down Syndrome Project, we're working with Emory University on that. The World Trade Center, we are looking at some outcome data to see if that had any impact on birth defects in our state.

Accutane I mentioned earlier. Water contaminants and neural tube defects, that was a national multi-state study. We have internally looked at our infant mortality data, coding and contribution to birth defects. In New Jersey, birth defects are not the number one cause of infant mortality, and we thought that was very odd, because in every other state it is. We found it to be a coding issue with our death certificates through this study.

We look at the accuracy of the birth defects reporting both on the electronic birth certificate and on the birth certificates in general. We are currently starting a pulse oximetry screening study using our data. We are going to be reporting on that next week at the CDC, at the birth defects meeting.

We have several programs, one registry. So again, the registry is this linchpin between several different projects. We have mandatory reporting from the screenings, 64 birthing facilities, a whole slew of medical professionals, and the case managements will also report back to us when they find things. A lot of times the hospitals don't identify somebody at birth, but the children will end up in case management services. So they will report back to us as well.

Again, we had a lot of different services. We do notify each and every family that we know where a child is living that has been reported to the registry. If we know the child is deceased, we don't tell the family that they've been registered, so as not to upset them. We provide the link. Case management coordinates with Part C, and we provide a whole different menu of health and social services with federal, state and local resources. We try to help the families as best we can.

Again, all children reported to the registry are directly referred out. I know I keep saying this over and over again, but this is really why we're there as a birth defects registry. We feel very strongly about our link to services. The case managers will assist families. They try to contact everybody within seven days of our getting the referral to them. Sometimes that's not feasible. Again, when it is an urgent need, they will contact them the same day. It is decentralized in each of the counties, and again, the medical home linkage is provided through the case management services.

Why does it work? We have law and rules for the birth defects registry, the Hearing Screening Program, and the Newborn Blood Spot Screening Program. They all have their own separate laws and set of rules that mandate not just the screening, but also the reporting to the registry. So we have our little stick in our hand.

We get funding from a variety of sources, and I hope I haven't forgotten anybody on this list. We used the block grant to help fund the registry for our case management services and our hearing project. HRSA funding, we have the Universal Newborn Hearing Screening Grant, and I know HRSA and MCHB are the same, but I wanted to separate that out. We have CDC funding for the surveillance. We are trying to develop an electronic reporting system for the birth defects registry, so we have a cooperative agreement for that, and also for our hearing screening project.

We have state and county money for the case management unit. The hospitals purchased the blood spot kit, and the fees generated from that fund the entire Newborn Blood Spot Screening Program. I think it is over $6 or $7 million that we generate in revenue from that blood spot screening.

We are part of an integrated system. We have been doing this for many, many, many years, so it really works very well. We communicate. We literally sit next to each other, we meet with each other on a regular basis to make sure that the communication in the linkages are made. The data is just part of the program. It is expected that we're there. We are not living somewhere in a different building with a whole group of people who don't understand the issues. The data folks understand the issues, work with the programs, so I think we have a better system than most people do.

We are integrated. We have partnerships with pretty much the gamut of folks who will touch the children, from Society Security to the Labor Department. We have a lot of buy-in from the agencies and the hospitals because we do provide this direct link to services. The agencies know that we're going to do that, and they feel that it is part of their obligation to then report to us to make sure that the kids do get the linkage. Otherwise, they would each have to have their own system in place for case managing these children.

It provides us the ability to meet a whole bunch of different challenges. We have public involvement through rule adoptions. We get a lot of referrals through the Governor's Office and from the Health Department Commissioner's Office. We respond to those. We have the data to do it, plus the services to assist the family with.

We have a lot of parent involvement on our advisory panels that we have. Again, we have early intervention and case management, which also have strong parent components as part of their process.

Challenges are of course funding, direct, and indirect. Money is always short. Confidentiality, we are always very concerned to protect the confidentiality of the families and the children that are registered

with us. So we are always on top of that. We are always educating new staff about confidentiality. We have to sign waivers of confidentiality, the whole nine yards.

Staffing is always an issue. When you are dealing with as many different birthing facilities, not to mention all the other health care providers in the state, staffing turnaround is tremendous. One of the reasons we do the annual visits to the hospitals for the birth defects registry, and we also do a separate annual visit for hearing screening, is to make sure that the staff are up to date, that they understand the reporting requirements, and they understand all of the components that they need to do.

Our audiologist that we have with the Newborn Hearing Program also goes out to all the hospitals. Right now she is focusing on the major audiology providers to make sure that if they say that they can test and screen children, that they actually can do it and have the proper equipment to do that.

Again, manual versus electronic reporting. Right now, everything for the birth defects is manual. Once we get the data, it is electronic. But getting the data is manual. We're really working on that to try to get into the 21st century, not to mention beyond. But HIPAA issues are primary, so we want to make sure that whatever we do doesn't violate any of the HIPAA statutes.

The benefits are it is cost effective and efficient. It is amazing at what you can do with a little bit of money and a few people when you have a system that is integrated and that you all work together. We do timely identify children and direct refer to case management. Most kids with the birth defects are registered by their sixth month of age, so we can make that link very quickly in life. It fosters a lot of communication, and it has built a good feeling with our partners. We have a good relationship with our hospital association, we have a good relationship with all these parent groups.

We do have data to answer public concerns if they are worried about any kind of cluster, or just in general they want data for research purposes, we have that available.

So the bottom line is we have a system for early identifying children. We have law, we have rules to provide the framework for that. We linked to services, and we find that that has really helped encourage reporting, and not just from the agencies, but also from the families involved in the system.

It is cost effective and efficient, and it ensures for the families that participate, coordinated care and linkage to a medical home. We do have a lot of quality control issues in place to make sure that the data that we're getting is accurate and reliable. We do work constantly to foster this idea of communication and team, not just the team at the state, but our philosophy is this entire care provision system is part of the team, and we want to work together with that.

I'd just like to thank everybody for funding us.

(Laughter.)

MS. BERES-SOCHKA: And hope to continue. If you need to contact me, there is my contact information. I hope I haven't talked too fast, which I have been told I tend to do.

DR. HOWELL: Thank you very much. I have two questions of you. The first is on your slide 24, you have a number that 113,000 had been screened.

MS. BERES-SOCHKA: Right.

DR. HOWELL: There were 3,458 abnormal results. Did that include the sickle?

MS. BERES-SOCHKA: No. Sickle cell was in addition to that.

DR. HOWELL: Right. Now, that's an interesting figure, because you basically have about 1 in 40 patients that you have screened that has an abnormal result.

MS. BERES-SOCHKA: Right.

DR. HOWELL: Which is a substantive figure. I know at our last meeting, the Mississippi data were reported. They had, as you recall, 1 in slightly over 300 patients and so forth.

Brad, can you tell us the percent -- oh, that's right. Brad's on his way to Hong Kong. I apologize. He will not hear us on his way to Hong Kong.

(Laughter.)

DR. HOWELL: But that is a very interesting figure.

MS. BERES-SOCHKA: Well, just to clarify, the sickle cell trait we don't do active follow-up to disposition. We provide the families with information that they have the trait. We give them some information. We let the physicians know, and then we don't follow them from there on out. This is fiscal year data, and again, we're still getting some numbers in, so it will go up.

DR. HOWELL: It's very interesting because obviously that's 3 percent of your population that has an abnormal result, which is a very interesting figure that obviously we are interested in identifying. Obviously many of those patients identified would be amenable to treatment and prevention of serious problems. So that is a very formidable figure.

Dr. Alexander?

DR. ALEXANDER: Is that abnormal result a lasting abnormal result? Or was it one that requires additional testing?

MS. BERES-SOCHKA: Those are the ones that require follow-up testing and screening.

DR. ALEXANDER: So it's not a final --

MS. BERES-SOCHKA: Well, this is not final. This is a provisional count. It is 271 now for fiscal year '04, but it will probably be in the five-hundreds by the time we get all the confirmatory testing done.

DR. HOWELL: The 500 would be a high figure, but it would not be terribly different from the data from Mississippi. But at 500, I have already calculated that you would have 500, and that would be 1 in 227 of your positives. But I think those data underline the importance of the expanded programs that you have in place and so forth.

One of the things of course that has been very tedious for the programs nationally that you have worked hard on in New Jersey is follow-up and identifying patients that have a screen-positive test in one of the areas that has been particularly complicated nationally has been hearing screening.

What are your data on the ability to identify persons that have a positive initial screening test? How successful have you been in following them?

MS. BERES-SOCHKA: I have to apologize. Somehow the slide that had the hearing screening data got deleted from my slide show, because I had the state data for last year. We are screening now

about 99 percent of all live births that occur in New Jersey. So screening, I think we have done as much as we can do.

The follow-up data, we have been working very actively with the hospitals on encouraging them to have follow-up programs that can actually talk to the families as opposed to just telling the doctor. We wanted them to reach out to the families themselves. We just broke the 50 percent mark that we know that the families have gone on for a second screen or a final diagnosis. So we have a lot of work to do.

We just started universal though in 2002, and we knew it would be at least a four or five-year program to get everybody onboard and doing it properly. We have emphasized from the beginning the importance of follow-up, but it takes a lot of education and hand holding. It is a different model for hospitals to follow. They are used to the blood spot screening where the state does the active follow-up. But the legislators in their wisdom wrote the law that the hospitals have to do the follow-up. So we're trying to work with the law that we have and the system that is in place to get everybody on board to up the numbers.

We are optimistic that this year's data will be in the 70s, if not higher. We've also reached out to all the pediatricians, we have the AAP chapter Champion Program which the American Academy of Pediatrics has identified, and each did a chapter champion for hearing. He has been doing grand rounds at all the hospitals. I think he is through 20 hospitals. He is 64 and he is doing this as a volunteer. So to get him to do all in one year was a little bit overwhelming for him.

He has also presented at the American Academy meetings that they've had in New Jersey state. We are doing a series of lunchtime visits. We have six maternal child health consortiums that the Health Department works directly with. The state is divided into six maternal and child health regions. We are working with those folks to get into doctor's offices directly. So my staff and the consortium staff will go into hospitals. We just had our first visit yesterday, so I'm not sure how it went since I'm here.

They were going to present the whole newborn screening, the hearing screening, and why it was so important to do follow-up, and what their role as a pediatrician was. That it was no longer acceptable to say wait until they are three or four, then we'll test them. The technology exists, and it is very important with all the research that you get the children into some sort of intervention by six months of age, and that they'll do as well as their age peers. So we're doing a lot of things on the follow-up front to try to up those numbers.

DR. HOWELL: I think your problem there is not unique, but I think that obviously we're extremely concerned when you have an abnormal newborn screening test, and then you are not able to identify the persons who had that abnormal test because you don't know whether -- in that 50 percent, you would assume that there might be as many patients who have an abnormal test as in the 50 percent you followed, which means that you are potentially missing half the customers. That is in deference to Dr. Edwards, half of the patients.

Are there other comments on this interesting data? We have comments everywhere. We'll start with Denise since you're right opposite me.

DR. DOUGHERTY: Okay. As you know, one of the issues with a lot of this early identification is whether the treatments actually work, and knowledge about what treatments and interventions people are getting. So I guess I would ask if you in your case management/EI process, you have a box that is periodic monitoring and follow-up.

I'm wondering if the effectiveness of the treatment and the parent satisfaction with the case management, which are two different aspects of quality of care, are being followed at all.

MS. BERES-SOCHKA: The case management units and early intervention have a very active parent component where they do needs assessment surveys and satisfaction surveys. It is my understanding, and this is not my personal data, but it is my understanding that the parents tend to be satisfied, but there is always room for improvement from that. What was your other question? I'm sorry. I dropped it.

DR. DOUGHERTY: It was about a systematic way to track what treatments children are getting, and whether they are improving as a result of those interventions.

MS. BERES-SOCHKA: Part of the new early intervention database system will be tracking those sorts of issues. So we'll have an electronic system in place to answer that question.

DR. HOWELL: Derek?

MR. ROBERTSON: Yes. First of all, thank you very much. I thought it was very impressive in general. What is your definition of final case disposition?

MS. BERES-SOCHKA: Diagnosis. When they get the final diagnosis. The confirmed diagnosis through whatever laboratory testing.

MR. ROBERTSON: And then do you make sure that the child is with a specialist?

MS. BERES-SOCHKA: Right. We have a whole cadre of specialists throughout the state. We have a consultant list broken down by specialty areas, so that when we identify a child with any one of the conditions, we know by our consultants. I have a copy if you'd like to see that list. So you can refer them directly to those consultants. They are throughout the state, so we try to make sure that no matter where you live, you can have access to a specialist.

MR. ROBERTSON: And does it go anything beyond that? I know like for my child recently got a letter from the state because he was school-age, and it said well, you are school-age, you are on your own. Keep going, keep doing the things you are doing kind of thing.

MS. BERES-SOCHKA: That's the advantage of our case management system. Because until the day they turn 22, our case management units will monitor and help the families manipulate the service system. As they approach 20, 21, we have a whole program in place for transition where you are teaching the families and the children themselves to advocate for the needs that they have and the services that they require.

But the case managers, we don't just throw them out the door once they pass early intervention or special ed. We keep them in case management until the day they turn 22. There is a whole process in place for transitioning them to adulthood. We work very closely with our Division of Developmental Disabilities, which is in the Department of Human Services, to make sure that all bases are covered.

MR. ROBERTSON: Thank you.

DR. HOWELL: Dr. Howse?

DR. HOWSE: Thank you. Leslie, that was a very impressive presentation.

It was so interesting to follow how the state has done its planning to go from one registry, and then to spin off in a very integrated way the diagnosis and the follow along, the referral services, and the family development. So thank you very much, very cogent. You talked a little fast, but it was a very cogent presentation.

MS. BERES-SOCHKA: I'm sorry. I apologize. I'm from the northeast.

DR. HOWSE: You did great, you did great. I have a very practical question. In the series of slides that dealt with the precise conditions that are in your newborn screening panel, how do those conditions relate to the recommendations in the ACMG report, question one.

Question two, what is your state policy with respect to reportable conditions? The report only screenings.

MS. BERES-SOCHKA: I'm sorry. The state only reportable?

DR. HOWSE: The conditions that are report only. The ones that can't be treated, but they can be detected.

MS. BERES-SOCHKA: Oh, okay. We do let the families know. The ones that are mandated by the state, we have this whole system in place. Once you get the positive, we tell the primary care provider, and the families are notified. It is the requirement of our law and our rules.

Even the ones that have no known treatment, we still have to tell the families. It is the right to know because you've done the test. And that was one of the things, and I was not on the advisory panel. My responsibilities are primarily birth defects and hearing, so I wasn't participating in that advisory panel. But I understand that a lot of their conversation about what to add and what you should add had to do with that issue. If you have a disorder that has no known treatment and no known case following, do you test for it, for one? And then what do you do with that? But in New Jersey, as long as it is mandated by our state law to be screened for, we will notify the families of the positive.

DR. HOWELL: Steve had a question or comment.

DR. EDWARDS: One of the things that I think that you have that is very pertinent to this committee is that you have gradually increased the numbers of tests that you've done over a three or four year period of time. Of course that is very germane to our work, because one of our recommendations would be a major increase in tests for many states.

But I wasn't clear from your discussion about how the individual patient that is screened gets into the case management system. Is that after they have screened, and then the screening has been reaffirmed that they get into case management?

MS. BERES-SOCHKA: Right.

DR. EDWARDS: Or does the case management come into play once they have been picked out in any way?

MS. BERES-SOCHKA: When they have been screened positive on their blood spot, through whatever the laboratory testing is, they get reported immediately through an electronic link up between the lab and our follow-up for the blood spot program.

They will contact the families and physicians and whatever. They don't report them to the birth defects registry until they have a final diagnosis. If there are other things going on in the family's life that requires case management, they will report. So it is a flexible system.

DR. EDWARDS: It's a separate system.

MS. BERES-SOCHKA: We look at the individual children and what they might need. But mostly it is when they get the confirmed test that they actually have that disorder that literally the person who got

the confirmation picks up one of my registration forms, walks over to my staff and hands it over. So that same day, we'll get them into the registry and refer them out to case management services.

DR. EDWARDS: And then the other question I was going to ask, as you have increased the numbers of conditions for which you screened, have you had to have a proportionate increase in the number of personnel that you hire?

MS. BERES-SOCHKA: Yes. That's why we rolled it out the way we did. To suddenly overnight add 20 to 30 new disorders would have overburdened the staff. They are not my staff, so I think there were four follow-up as well as clerical staff and data people. To suddenly dump 30 more disorders on them overnight would have grossly overburdened the system.

The complexity of some of these conditions is such that we wanted to make sure that as we rolled out the condition, that we had trained staff, we had identified consultants who would be available to help the families. We wanted to do it in such a way that we didn't hurt the families by giving them false information or just by the sheer, when you are doing so much inevitably something gets dropped. So we thought it would be better to stage it, add a few more staff, get them up and running, add a few more conditions, add some more staff and so on.

Again, this year we are going to look at the balance of what can be screened for, and the advisory panel will make a recommendation to the Health Department and the Governor as to what they think we should add. At that point, we'll look at the fiscal side of it as how many follow-up staff will we need to handle that additional number of people, and we'll plan accordingly.

DR. HOWELL: Bill?

DR. BECKER: Thanks, Leslie. That was a fantastic presentation, the program to be modeled after.

You partially answered one of my questions. It has been a contention of mine for awhile now that the demographic information on a newborn screening card is oftentimes the first entry that an infant will have into one of the state's various databases. But it is not as complete as certainly the vital statistics record and probably doesn't include many of the elements for even the birth defects registry or some of the other registries that you've integrated.

Does that create a problem for you guys to have a limited demographic data set on an infant who you know needs case management or early intervention services, or do you just create a placeholder, start the case, and try to work the vital statistics information in and all that other stuff later?

MS. BERES-SOCHKA: There's no one answer for that. The laboratory gets the card.

DR. BECKER: Right.

MS. BERES-SOCHKA: They do some of the data entry by themselves, so they currently are not linked to the electronic birth certificate. Because of the timing and urgency of getting that blood spot done, the electronic birth certificate comes to the Health Department at a minimum of every seven days from each facility.

So if you waited for that, you'd be in serious trouble with getting the results done. So there is that issue. So you are kind of creating two databases. I know one of the long-term projects, and I can't speak whether it has been done exactly as they were talking about, somehow linking those two to make sure that what you had with our blood card eventually got linked to what is in the electronic birth certificate system.

Again, that is not my area, so I'm not exactly sure if it happened, or when the plan is for that to happen. As far as getting them to case management services, we will call the birth hospitals at the registry to complete the form if our blood spot program doesn't have the information. But typically by the time the follow-up program has that data, they at least have the parent's name, phone number, and address that is accurate, so they can get the information from the parent on the telephone.

Again, if they don't, the birth defect registry staff will call the birthing facility to make sure that we have that data if it is not on the version of the EBC that we have at the time. So it takes a lot more work, but again, our primary focus is the kids and making sure that they get what they need in the area where they live.

DR. BECKER: This is one of the challenges that the states have with these different databases in trying to either create what the IT guys call a common portal, or continue to require duplicative entries being made, and very similar demographic information. Name, date of birth, mother's name, on maybe four, five, six, seven different pieces of paper all coming out of the hospital. Can we somehow simplify that? But then on the other end you've got the state trying to figure out how to integrate those databases. That is a challenge.

My other question is a very specific one. It is something I'm trying to get started in Ohio actually. You made a statement that you guys do direct referrals to your specialists. Are those referrals I guess I'll say official referrals for the purposes of medical care, providing medical care? My real question, my intent of this question is how do the payers, Medicaid and insurers, deal with that referral process? Can the state refer cases to the specialists, and how is that handled?

MS. BERES-SOCHKA: Well, it is up to the primary care provider to do the actual insurance paperwork and whatnot. I can't really answer that question. Health reimbursement issues are not in my field. I'm afraid if I say anything, I will misspeak. But I can get you the information. I'll talk to our case managers and our follow-up program. If you give me your email, I'll let you know the answer. That's a very important question, but I don't want to say something and then have to call the committee up and say please strike that from the record.

DR. BECKER: Right. Perfect. Thanks.

DR. HOWELL: We have a comment from Piero, and Coleen is going to have the last word.

DR. RINALDO: As Dr. Edwards mentioned earlier, this is a bubbling program with a lot of additions, and clearly metabolic disorders are becoming an important part of it.

My attention was one of your slides where you actually are proud of the fact that you have multiple programs in one registry. So I am aware that New Jersey is one of the few states with a law that actually mandates metabolic postmortem screening, especially when there is an opportunity to link data from the newborn screening program with the postmortem results. It clearly seems pretty obvious to me.

So do you have any plans to involve the medical examiner's office and actually incorporate those data in your registry?

MS. BERES-SOCHKA: The medical examiners are obligated by state law to report any abnormality, congenital defect or whatever to the birth defects registry. We find reporting from them is spotty. One of our initiatives has been to reach out to them individually.

Unfortunately we don't have the same stick that we have with the hospitals with our hospital licensing rules. We would have to report them to a different board over which the Health Department doesn't really have the same jurisdiction.

So we are working with them, but it is going to be a slower process to make sure we get everything in. However, we do review all the death certificates for children who die under the age of three. So we pick up a lot of the information that way. It is kind of the

back-door approach to getting information on what the children may have had at the time of death. We will also ask for autopsy reports from the birthing facilities if they can get them for us. So it is not really yes, but not no either.

DR. HOWELL: Coleen?

DR. BOYLE: Leslie, thanks for the overview. It was terrific.

I just had a question. Yesterday we talked a lot about long-term follow-up, and actually trying to understand the impact of treatment and the natural history of the disorder. I wasn't quite sure with your system whether you would actually have that data available, or you'd actually have to go back to the individual service provider level to get that.

MS. BERES-SOCHKA: Right. That's what we're using your money for. One of the weaknesses of the birth defect registry, it tends to be a snapshot in time with some updates of conditions, but we don't have the electronic ability right now to follow them long term.

The hearing screening log gives us that ability. The registry is being set up to track them over time. With the birth defects, it was kind of a picture of time. We're trying to develop an electronic link out to the case management unit so we can take advantage of the system that they have of tracking and managing the children so that we know one, if we refer John Smith, that John Smith actually received treatment or did not receive treatment, and if they did, what did they receive? How many things, and over what time frame?

So we are at the preliminary stage of that project development. We now have the technology in place, and the computer folks, programmers and what not, and we're working very closely with case management and my staff to make sure that the things that we're putting in that system can answer those questions. It is a ways away, but we are trying to address it.

DR. BOYLE: So that would include all the children in the registry, not just those with congenital malformations, but those with metabolic disorders?

MS. BERES-SOCHKA: Right.

DR. BOYLE: Okay.

MS. BERES-SOCHKA: We would like to follow all of them through the case management system, which is what we are able to do.

DR. BOYLE: And a last quick question. Do you have a sense of how many children in that period from birth to 21 actually avail themselves of the services?

MS. BERES-SOCHKA: I had that number. It is in the 10,000 to 20,000 a year avail themselves to the services.

DR. BOYLE: What percentage of all children?

MS. BERES-SOCHKA: Probably about 50 to 60 percent, but I'm guessing. I don't have that. That is one of the things when we develop the data system, we'll be able to answer directly. We do

know that 90 percent of the referrals come because of the registry sending the referral out to the case management unit.

It is an interesting process in case management. The children tend to come and go over time. There will be a lot of initial heavy use because they are trying to find services, or they are trying to find financial assistance. The case manager will work with them to make all those links.

They might disappear for a few years because the families feel competent at advocating for themselves, or they have what they need in place. Then you might have some trigger event where they need more, so then they'll come back to case management.

We also have families who up front don't want any help, they think that they can handle it on their own, and many can. But then maybe five, ten years out, they hit a brick wall. They lose insurance, they lose coverage or whatever. They'll find the letter that we sent them from the birth defects registry and we'll get a phone call. They'll say, we have this letter that you sent us when the baby was born, do you still do this? We will make that link at that time. So it is a system that is always in transition, and we try not to lose or forget anybody.

DR. HOWELL: Leslie, thank you very much.

I'm going to follow up one brief comment on a question that Denise asked that I think is critical. That is does the treatment that is being recommended make any difference?

One of the things that we're going to be certainly thinking a lot about is there certainly is a changing interest in newborn screening to include conditions that don't clearly have what we would call traditional treatments such as a diet or a vitamin or something.

One of those that is of great interest nationally is Fragile X. Again, research efforts are currently undergoing right now to see whether or not certain programs indeed do make a difference. I think those are going to be very important to know that yes indeed they do make a difference if started at this period of time and the long-term effects are positive, which I think will weigh heavily on the decision of detecting those conditions. I think it is very important to know the best we can whether what we are doing is really making a difference. Sometimes things that we think are working don't necessarily do what we think.

It is time for our break, and we actually are a bit over our time. So we'll be a bit tardy returning, but not much. Let's try to get back by 10 of.

(Recess.)

DR. HOWELL: One of the discussions that has surrounded the committee for some time and that people have been thinking about are the formation and importance of subcommittees that would focus in various areas of importance. During the course of these discussions, there have been a number of areas that come up.

Since this is a federally established committee that operates under very specific rules, I've asked Dr. van Dyck if he would be good enough to spend some time with us talking about subcommittees, how they are formed, how they must be formed, and so forth, and how they work. So as we proceed to identify areas of interest and so forth, that we work within the established federal guidelines in this area.

Peter, can you walk us through that, please?

DR. VAN DYCK: I'd be glad to. It is really not very complicated. I think this is easy. The chair of a committee has the discretion to establish subcommittees as he or she pleases,

generally. Subcommittees are established with members of the committee. The chair appoints a chair of a subcommittee, and usually the interest of people who volunteer for those committees may assign then committee members to those subcommittees, and has the ability clearly to place people if they choose not to participate, or if they'd like a different committee.

So the chair really has the ultimate responsibility for establishing the subcommittees, and then staffing them and naming a chair. Subcommittees do their work both between meetings and during meetings. So a subcommittee can get an assignment, work on it usually by phone, conference call, which the staff will be happy to set up, coordinate and facilitate, and do work in between meetings, then can come and report out at the meetings and get general consensus from the entire committee.

The staff will be happy within reason to help on research or those kinds of things that subcommittees need in between times to help in the development of that material, or to help write certain things. If a subcommittee needs to meet face to face over a very difficult issue, or for some other reason, then we can facilitate and pay for that subcommittee meeting.

Clearly we'd prefer not to do that if we don't have to, but we can on issues that are really necessary to meet as a subcommittee face to face.

The work of the subcommittee, regular minutes need to be taken of those meetings, and certainly when subcommittees report, that is part of the official record.

There may be, at the discretion of the chair, besides the committee members populating a subcommittee, at the invitation of the chair, there may be others who are not members of the committee from the community who could be on those committees or contribute special expertise, or be asked on an ad hoc basis to come present to a subcommittee, or to prepare material for a subcommittee, or to even sit on a subcommittee if the chair would choose to do that. So that is another option that the chair has.

Unless there are questions, I think --

DR. HOWELL: There seem to be questions.

Denise?

DR. DOUGHERTY: If, say, a subcommittee decided there was a need for a consultant who had to be paid to write a paper or something like that, do some analysis, could that be paid out of the advisory committee budget?

DR. VAN DYCK: That can, at the discretion of the chair and the staff, yes.

DR. HOWELL: And I would think that that could be invaluable in certain circumstances.

DR. BOYLE: I was just going to say that on at least one other advisory committee I have sat on, one other rule that I didn't hear mentioned, but you alluded to it, was that a subcommittee had to have at least two members of the full committee on there, and it could have a number of other members as well who are not full committee members. But you have to have two.

DR. VAN DYCK: We have no rule related to that. I would hope that there would be more than that from the parent committee on the subcommittee. I would hope that it would be populated mostly by people from the committee.

DR. DOUGHERTY: Looking at the liaisons, they are included as well?

DR. VAN DYCK: Yes.

DR. DOUGHERTY: Okay.

DR. HOWELL: Are there any other questions about the general requirements of how these subcommittees are formed or anything?

(No response.)

DR. HOWELL: It seems to me that there are several issues. One is that although the members and so forth are appointed by me, I would want to rely essentially totally on the membership and your expressed interest and expertise. Also, I think that we should come up with an idea of how many subcommittees we should have, and what they should address.

There are an enormous number of issues. I would think it would be highly desirable if we had two or three, and I would think three is the magic number of members of this committee on each of the subcommittees. Obviously as you look around the table, with the size of the committee, which is rather small, that means that we can't reasonably have a huge number of these subcommittees to begin with certainly. So we should come up with some finite number of subcommittees that have a key focus that we've identified, and try to identify those.

Let me tell you what I would hope we might do today. Let's be specific and practical. It is to come up with the subcommittees that this group thinks would be most helpful in moving the project along and so forth. And then I would like to have from you by lunch today an expression of your interest in the committee. I would prefer having that as a little written thing so that you don't just kind of say I'd like to be on that, because I'll promptly forget. So if you could just let me have after we are through here as we are going to lunch, and then we can fairly promptly look at this list and try to get a group together so that before we leave today we can have some ideas of what the subcommittees should be, and who the core people are.

It is my impression that the subcommittees are responsible for forming their own agenda and work plan. But this overall committee ought to hear about that overall agenda before a group goes off. So it seems to me that the committees, if we could identify them today, they could be fundamentally established in some way. The group could then have conversations and so forth in the coming weeks, develop a work plan or an agenda, whatever you want to call it, and then at the next meeting we can have as one of our agenda items for the big meeting, the spokesperson for each of these subcommittees discussing what the agendas might be, and then we can proceed at that point. Does that seem reasonable to the group? And is that consistent with the guidelines, Peter?

DR. VAN DYCK: Yes.

DR. HOWELL: Does that make sense to people?

Piero?

DR. RINALDO: Well, it does, at least to me. One point, I think we first need to have a better idea of exact definitions of the subcommittees. I would like to pick up where we left yesterday with four major areas. I don't know if it is HRSA or whose pyramid that was. But I really think that Infrastructure Services, Population Based Services, Enabling Services, and Direct Services certainly are a good place to start. I don't know what the rest of the committee thinks.

DR. BECKER: Rod, can I ask a question about setting a subcommittee agenda process that you just outlined?

If we assemble and assign some subcommittee groups today, and they can certainly work I guess to formulate their agendas, if we wait until April to submit them for overall review, it means really that no subcommittee will work. Well, I guess that would be working on their agendas between now and the middle part of April.

Is there an alternative way perhaps of moving their agendas along from formal committee review? I'm just asking.

DR. VAN DYCK: Yes, there is. If the chair decides that the subcommittees within 30 days form an agenda, then the staff can make sure that it is done electronically. The staff can email those out to everybody, and there can be a comment period of a week or something. If it comes back, the chair can then decide through the electronic mechanism that the agendas are okay and email you back that it is okay to proceed. So that can be done electronically.

DR. HOWELL: Denise?

DR. DOUGHERTY: I like Piero's idea of those four levels. But I wonder if we might step back and think about now that the report will be out and states would be encouraged to adopt this new panel, what challenges are they going to face in adopting those? How might we develop some recommendations that could assist, sort of taking it from that perspective?

So I have a list using that kind of thinking, and my reference to you is that it included two things from the infrastructure, which was IT, which I think is an opportunity with the current Secretary, David Brailer and so forth, and financing, to come up with some financing alternatives.

Then I guess as a longer term issue, I'd like us to have committees on the criteria for additional selection of tests and start thinking about quality improvement. I think, and others can correct me if I'm wrong, that these IT linkages to start getting some data collected and analyzed, plus financing so that states can start actually implementing these tests would be two critical areas. Medicaid is supposed to be reduced this year, and block grants may be reduced. So I think we really do need to help the states look at some alternative financing schemes. So those are both in the infrastructure. I thought I'd raise there. There are other important issues.

DR. HOWELL: And Jennifer?

DR. HOWSE: I'd just like to build on what Piero and Denise have suggested, and add another area for consideration, which would be education and training, professional training and parent education.

I know as far as my own organization is concerned that that is an area that is quite meaty. There is a lot of work to do, both on the consumer outreach side, as well as on the professional support and training for professionals. So that would be an area that we feel is quite important.

I really like what Denise said about being implementation-oriented in the way that the subcommittees would work. That our job with the report moving into the public domain for comment, it is out there, it will be out there. Then our job as a committee would really be to be supportive in very specific ways to states in their expansion of their newborn screening programs along the lines that are recommended in the report. So I'd just like to add that area of education and training as a candidate for a specific subcommittee.

DR. RINALDO: Actually, I think we are probably calling the same things with different names. If you look at the detailed list, under population-based services, the only bullet is education. So if there is a subcommittee on population, there are services, there will be, by all means, a subcommittee on

education. So I think we are all saying the same thing. It is just a matter of how we call them. To me, it is more of a concern of how many of them. So it depends on how much we define the focus.

DR. DOUGHERTY: It might be helpful to talk about how long the subcommittees would have to do their work, and what we would expect the subcommittees to come up with.

DR. VAN DYCK: Certainly a subcommittee is not in perpetuity. So a subcommittee has a task, it has a function, it submits a report. The report gets voted on and accepted, and the subcommittee can disband, and presumably a new subcommittee would be established. So there is some sense to choosing those items first that are the highest priority clearly, and then moving onto secondary or less priority issues.

DR. HOWELL: And I would think that in certain areas, such as the one Jennifer mentioned about education and training, that will probably have specific goals at the outset that will be accomplished. But I would imagine that that committee's work would indeed be here in perpetuity. I think that there will probably never be any end, but there will be new projects within it. I think that they may have to continue.

Let's hear from other folks about some of the subcommittees. I think that these have been excellent suggestions that we've heard, and so forth.

Joseph?

DR. TELFAIR: I would second Peter's comment, because that is one of the things that I was going to bring up related to just the temporal aspects of the subcommittees. It seems to me that there is in terms of priorities, that's one thing. But the other piece is also in terms of what needs to come before the other.

I mean, there is probably some work that needs to be done in terms of defining what would go into a subcommittee, for example, on the issue of counting prior to you also talking about the issue of subsequent training and that sort of thing. So I think when we talk about subcommittees, we also need to think about in terms of priorities, we need to think about what needs to come first, and what would be the most logical thing. Or have that as part of the committee structure, what it would work on, setting a time frame, and that sort of thing.

DR. HOWELL: Right, right.

Coleen?

DR. BOYLE: I guess I would agree with everything everyone has said so far. I like the list that Peter presented yesterday. I thought that was pretty comprehensive. I think it actually has some of the financing and reimbursement issues in it. I would agree with what Joseph just said that I think we can take this list and sort of prioritize what we think would be most appropriately acted on first, which is if we went through policy development, clearly I would think laboratory standards would be the number one priority under that in terms of the implementation stage. So maybe a committee could actually be working on developing a uniform system or uniform reporting, laboratory standards there. Maybe it would help if we just went through that list and decided which was most implementable.

DR. HOWELL: Coleen is talking about the list that is in Tab 11, in case you don't have it. It is after the slide set there and so forth.

Coleen, you're suggesting that we actually go down the list here and so forth.

Would someone like to look down the list and see some of the things that pop out at them? The ones that are highlighted, as you know, are the ones that we have heard formal reports prior to the committee and don't necessarily have a prioritization and so forth.

Derek?

MR. ROBERTSON: I don't know if this was said before, but I think follow-up is a major thing, follow-up protocols. It is one thing to do the testing and have it tested, and then we don't want to be losing the kids to follow-up. Exactly what is long-term follow-up versus short term. I think that that would be something significant.

DR. HOWELL: Amy?

DR. BROWER: And I agree with that. I would recommend we do go through this list and try to prioritize it. I think that as some members of the committee focus on policies and definitions, there is also given our expertise some members who can work on future things like new technologies, new pipelines and new tests. So while those aren't as pressing temporally as getting the ACMG recommendations and panel in place, I think working ahead of technologies and new tests would be important as well.

DR. HOWELL: Let's look at the list. We've had several folks, and the first thing is Infrastructure Services. That has many subsets under it. Policy Development. In that first area, I won't go through each of the little things here, but is there one or more of the items that should be a subcommittee in that first thing under laboratory standards?

DR. COGGINS: We've had the variation in the test panels state by state, and we've made some recommendations on that. But then the implementation of that, in particular the application of different cutoff values and different test protocols state by state. So I think that might be an important one to pick up on and prioritize that.

DR. HOWELL: And how would you entitle such a subcommittee that would give a little meat to its charge?

DR. COGGINS: Well, I think it comes under this laboratory standards. It is just a piece of that. But I think that is a high priority within laboratory standards.

DR. HOWELL: So we're having a suggestion that there be a subcommittee formed that would have laboratory standards as is charged, and that would look under that at some of the things that are listed here under Peter's bullets here, such as cutoff values and things of that nature.

DR. VAN DYCK: I'm wondering if we might want to do something one level higher. Is the subcommittee perhaps a subcommittee around policy development? And then it is the subcommittee's job to prioritize the items that they want to do in some kind of order within that area and bring it back to the full committee for approval, rather than choosing individual things within the major bullets. Only as a suggestion.

DR. HOWELL: Any thoughts about that? Moving it up would take the residual blood spot, the follow-up protocols and so forth, all in that first thing.

Derek?

MR. ROBERTSON: I just had a question for Dr. van Dyck. When you say policy development, doesn't that involve almost everything in some sort of policy, even going down the line to system

development of integrating your systems? I was wondering if that was broader as to what your thoughts were in terms of how you were distinguishing policy development from some other thing.

DR. VAN DYCK: We're distinguishing it here in actually being very specific. So if you're talking about policies for cutoffs, you're really talking about developing cutoff numbers. If you're doing follow-up protocols, then specifically you are recommending or working on the development of very specific types of protocols that could be used for policy development in states.

MR. ROBERTSON: You would be developing a policy -- for example, under your informatics infrastructure, would there be a policy on how you integrate public health and personal health services?

DR. VAN DYCK: Well, there could be, and that could be then one of the recommendations from a subcommittee that's working on informatics, is what pieces of their recommendations are policy, what are infrastructure development, what are definitions, so that there might be multiple recommendations that could include policy development.

And again, these aren't meant to be so tight that they only can fall within one area. Things overlap obviously. It is just meant to try and get the ideas down on paper.

DR. HOWELL: Any comments then about this first group?

(No response.)

DR. HOWELL: Peter, do you want to comment further about this? As far as you're concerned, the committee that would deal with laboratory standards as well as follow-up and the whole thing, would that be too big of a charge to consider, or not?

DR. BOYLE: I'm not sure about the overall scope, but I think within the laboratory standards piece, I think that's important. I would say, you know, because of the discussion we were having yesterday about the different test protocols and the different cutoffs that are in use, then that has to be a high priority within that. I would just like to see that covered somewhere under laboratory standards.

I agree with what Peter said, though, that I think maybe we should look at a higher level and set the agenda underneath that in terms of what can be covered, but just prioritize that.

DR. BOYLE: I was just going to say I think there are going to be different people, like we heard yesterday in the laboratorians versus public health folks in terms of a follow-up. I mean, it does involve different disciplines. Maybe that's okay. But if we are only going to have a few people on these subcommittees, I don't know exactly what the impact of that might have on our decisions.

DR. HOWELL: You're bringing up the prospect that there be a subcommittee that is focusing really on laboratory related things. It might include the storage of the blood spots or something, but not necessarily be the follow-up protocol people, which is a public health group you think would be a slightly perhaps different constitution.

DR. BOYLE: Well, if we're thinking about actually developing policy around protocols and follow-up and model things, I think that might be.

DR. HOWELL: Piero?

DR. RINALDO: I think perhaps part of the problem is that policy development is a very generic and broad term, because all these bullets under policy development really seems to be related to testing. So perhaps we can also look at the traditional components of a newborn screening program, and

then see if we should try to merge these various topics with sort of the traditional areas of education and follow-up treatment, evaluation, and testing.

So these are testing issues. I don't know. Maybe testing development, I don't know. Maybe the word "testing" somehow should appear in the definition of a subcommittee so that it is clear that we are talking about that component.

DR. HOWELL: Would you like to suggest a name?

DR. RINALDO: I'm thinking about it. Laboratory standards is close.

DR. HOWELL: Amy?

DR. BROWER: And I agree with what Piero said. I think we need to also include something on best practices or something that addresses not only the tests we're considering today and want to define laboratory standards around, but the next test. So we need a committee or an item on the subcommittee's agenda somewhere to think about best practice tests and next tests.

DR. HOWELL: Come up with a name of a subcommittee that would embody the things that you're talking about then.

DR. BROWER: I think the expertise is different between a laboratory test, analytical, specificity and validity, and another group that would do clinical utility. So I think those are two different sets of people or groups.

DR. HOWELL: Having said that, let's have some recommendations about how we move forward from that very thoughtful comment.

DR. VAN DYCK: Can you call it Laboratory Standards and Procedures? I mean, something that specific.

DR. RINALDO: Laboratory Standards and Procedures?

DR. HOWELL: That potentially could have within it looking at evolving technology, too. It could. Evolving technology should involve laboratory people.

DR. COGGINS: Yes. I think it should include looking at new technologies as well, and getting that encompassed within whatever we decide to call this.

DR. HOWELL: Well, is there a general agreement that there be a committee then, a subcommittee, that could be, for want of a better term, called Laboratory Standards and Procedures, and the group can decide what it wants to do. But we would anticipate it would look at the things that are listed here as testing strategies and things of that nature at the current time, but it would also seem very prudent to consider not only what we're doing, but some of the evolving technologies. The expertise should be able to look at that and so forth. So that might be one subcommittee. Does that make sense?

DR. EDWARDS: Yes. I think it does, and I think it fits all of the categories actually on there, not just the ones under the laboratory standards there. I think actually everything under that policy development could fit under Laboratory Standards and Procedures.

DR. HOWELL: I think it could. Well, is there a general agreement then that we would have a subcommittee of that type, and that that group should look at considering as Stephen suggested, all of

the things under policy development because they could at least have some insight into that and so forth. So I think that we have one subcommittee on the table at the current time. What about another?

MR. ROBERTSON: Sorry, Mr. Chairman. Just the only thing that I would want some clarification on is a follow-up protocol as it fits under those bullets there. I was thinking that follow-up is something separate from the laboratory. I mean, the policy development, yes. But I'm thinking it might be worth it to tease out follow-up completely separate. But maybe I'm wrong.

DR. HOWELL: Joseph?

DR. TELFAIR: I do think that, and I understand Derek's question, I hope, and Derek can correct me if I'm wrong. But actually follow-up can go in more than one place here. There is the overlapping issue of follow-up as it relates to the laboratory procedures, and then there is follow-up in terms of the actual work to be done related with the families. That would go in another category. So follow-up can actually go in more than one place. In terms of just logistically, it does go in more than one place in practice.

DR. HOWELL: I agree. It seems to me it certainly would not be the exclusive purview of this, but it seems to me it might be elsewhere. Certainly laboratory standards, procedures, cutoffs and so forth would be one of the high priority items once the recommendations are on the street for the expansion and so forth.

DR. BROWER: And I think, just to pick up on what Derek said, I think follow-up goes hand in hand with clinical utility. So which disease or disorder are you going to test for, and then does treatment make a difference, and does follow-up make a difference?

So I think somewhere in that follow-up we need to also include clinical utility and something about the new test, because that is how we evaluate them is to be able to look at the natural history of the disease, the current treatments, and strategies.

DR. HOWELL: Yes, Piero?

DR. RINALDO: I'd like to suggest a list of four possible, and then again, if you think it is worth it. I think one should be Laboratory Standards and Procedures, another one should be Follow-Up and Treatment, another one, and following to what was said before, I really think we should focus on Education and Future Additions that I think is really important to maintain this process defined as a constant loop of evaluation. And finally, one should combine IT and Financing. A little different, but again, it is --

DR. LLOYD-PURYEAR: What was the third one?

DR. RINALDO: The third one, Education and Future Additions.

DR. LLOYD-PURYEAR: Education and Future Additions?

DR. RINALDO: Yes. Future additions to screening programs. So new conditions, new technology. I think it implies the application of new technologies, new conditions.

DR. BOYLE: I guess Piero the last two, I feel like those are mixing apples and oranges there.

DR. RINALDO: I know, but I think --

DR. BOYLE: I mean, we don't need to handle everything right away.

DR. RINALDO: Okay.

DR. BOYLE: So maybe that's the way to do it.

DR. RINALDO: But I was trying to follow, you know, the concept that if each committee should have three of us means that realistically we cannot have more than four. That means some apples and oranges has to be put together.

You might decide at the beginning you'll only deal with IT issues, and then later it might go to financing. I don't know. I think it just is an attempt not to leave out important things. Am I the only one that feels that perhaps there shouldn't be more than four?

DR. HOWELL: No, I think it would be difficult to have more than four with the size of the committee. I mean, this committee, because the subcommittees need to be populated largely with this group, so the numbers don't work for more than four. But I think we should think about the areas.

Coleen, I actually think it would be great if we could do everything at once, which is my commitment. But on the other hand, we probably can't. But we ought to look at the things that are going to be the most urgent at the current time. I think education is going to be up there, because the public education, we were talking this morning some of us in the pediatric community about the importance of educating pediatricians, and obviously that goes out. But the education thing is going to be something that is going to be very important early in the game, I believe. Maybe no one else agrees except Jennifer. I know she does.

DR. RINALDO: And actually, exactly for that reason, I think the lesson we are learning here is that we are constantly in a catch-up mode. That's why I think linking education with future expansions is perhaps a way to prevent a repeat. Think of it. We are doing now testing for certain new conditions that have been in the book for 15 years. Now we are here talking about how can we reach out to our colleagues and professionals who have never heard of it?

So personally I think there should be a close link between education, not only retrospectively, but also looking forward. Whenever we seriously consider something should eventually join the newborn screening panel or a recommended panel, then education should start right away.

DR. HOWELL: Jennifer?

DR. HOWSE: Again, I'm going to just sort of come back to what Denise said about how can the committee think about being the most helpful and the most supportive to states as they proceed down what will be a long road to expanding their newborn screening programs. I mean, our situation is one in which the recommendations that are soon going to be in the public domain, you know, there is still less than 30 percent of the kids in the country who are even getting the recommended tests right now.

There are all kinds of issues around lab standards and procedures. So I guess I'm in practical mode with Denise. Not to look away from the future, certainly not that. But I guess I'm in practical mode with Denise about how we should assign priorities, and then from that derive subcommittees, at least for the first phase of the work. So I really like the subcommittee on Lab Standards and Procedures. I think that committee can also deal with new technologies that can be an aspect of the work.

I really vote strongly for a subcommittee on Education and Training for both health professionals, as well as for parents and consumers. I think there is wonderful information that is all scattered around here and there, and we could probably do some very useful and important work there.

Then this hasn't exactly been put together this way, but I sort of wonder what you all thought about a third subcommittee that really dealt explicitly with treatment and follow-up. It is a little bit of a different way to cut the pyramid. But if we sort of went from Lab Standards and Procedures, you know, how the job gets done and what are some of the new outlooks, Education and Training, how do we make sure that the people who are involved in this, whether they're parents or professionals, have the most contemporaneous information and resources? They know about resources. And then thirdly, we look at treatment and follow-up, so that as the conditions are identified, we follow best practices and programs that assure that the kids get the best treatment, that the follow-up is in place, and also that emerging treatment and exciting new opportunities for treatment also get integrated into that discussion.

So I would really see the three subcommittees working very closely in the feedback loop, just as you recommended earlier. I guess I'm sort of inclined to three rather than four, just because we're a small committee and we might need more than three people on a subcommittee to really help drive it. So those are my thoughts on it.

DR. HOWELL: Piero, why don't you, and then Joseph.

DR. RINALDO: I agree. Actually, looking at the list, perhaps the fourth one could be Evaluation. It is certainly highlighted as one of the bullets, and that actually has a lot of latitude. But I think we need to start talking about okay, a lot of things are happening, and trying to assess how effective they are.

DR. HOWELL: Joseph?

DR. TELFAIR: Yes, I guess I would kind of go back even before you started naming committees. I would just reiterate something. It seemed to me that for this particular committee, there is a clear set of goals and objectives that were set out, or expectations of what this committee would work on and do.

With that in mind, and then also there is a time frame to put this together. I guess I would recommend instead of just sort of setting a number of subcommittees to exist, it seems to me that a more important thing to do would be set what are the issues that this committee can expect to address, prioritize that list, do four or whatever committees at a time, knowing that one committee ends and another one starts up again. Instead of just saying we're going to do four, maybe you don't do four at one time or whatever. But I think still there is sort of future work, not sort of you don't have to do everything right now, but there is future work. So I would look at the list here, if that is what you are using as a structure, and think of it that way.

I think I'm hearing a bit of the conversation to put groups together or put work tasks or work type of groups together into categories, when it seems to me that one of the things to do is think about what needs to be done first, and then you can categorize them. If it is more than four, then you know that you have to prioritize and get things done organizationally and look for a scope of work or plan of work. That may be a better way to go, so I'm just suggesting that for the group to consider.

DR. HOWELL: I think the idea of looking at priorities has been driving the discussion about the subcommittees. I think your point is well made. To go back to Coleen's comment, we can't do everything at once. So I think that we do need to start off with the things that we think are the highest priority at the current time. We have much conversation. Let's start over here.

Peter, and then we'll go over to Denise.

DR. VAN DYCK: Well, I think, Joseph, that's kind of what we're doing by naming the subcommittees. I mean, I agree with you. I think that's the way to proceed. It seems to me that the last three that were just discussed, Lab Standards and Procedures, Education and Training, and Treatment

and Follow-Up, at least in my estimation would surface as the higher priority areas out of these lists. Financing had also come up several times.

Jennifer, I was going to ask you if Financing you think of in Treatment and Follow-Up, or do you think that's a separate area? Or do you think that rises to the level of a fourth committee? I also agree with you that three probably is -- it is hard to do more than three. It is possible to do four, but I think it is hard to do more than three.

DR. HOWELL: We have Bill, Denise, and Greg.

DR. BECKER: Yes, I agree with that. I like the suggestion that Jennifer has put on the table. I would think that what could happen next is if we agree to that sort of structuring and names to our subcommittees is then take the elements out of Peter's outline and let the subcommittees form their agendas out of those elements. Then at the chair's discretion, if one subcommittee doesn't address something that is felt to be particularly important or the committee points it out and it is felt important to be addressed at this time by that subcommittee, then the chair or the committee would direct the subcommittee to add those in.

So to a certain extent, the naming of the subcommittees, if we can agree on the general categories that are before us, then as they are assigned, the people who are assigned to work on these subcommittees thinking about the members that will be on those subcommittees, who they might want to invite to also participate on those subcommittees, then go to this template document and put their agendas together and bring them back for review and comment.

DR. HOWELL: I agree with Bill. I think that we are trying to get, or at least what I would hope we're trying to get is to get some general formatting. But the specifics of the work should really come out of the committees.

Let's have Denise and Greg who have been waiting, and then Coleen has come to life over here, and you're next.

(Laughter.)

DR. HOWELL: Oh, she has been very lively, but she has come to life again.

DR. DOUGHERTY: I guess I have two things. One is I have the same question as Peter for Jennifer, whether Financing needs to be a separate committee, though I agree three should be -- actually I have three things.

The second one was on evaluation. I think that every committee should consider evaluation as a cross-cutting issue. You know, you need to decide where do we want to get to, and then how will we know whether we got there as part of the committee discussion.

The third thing, though, is, and I wonder if maybe we shouldn't call it a subcommittee, but a low level or process for the whole committee. But I really think given the difficulties we've had with the deciding about the ACMG report that I think it is really essential for this committee to address what criteria should be used for the committee to recommend future tests.

We really need to set a process for that so that we don't have what we had, even continuing into this meeting. I really think the committee as a whole needs to grapple with that, but somebody probably needs to come forward with a proposal for how to do it so that the committee can comment on it.

DR. RINALDO: A quick comment. It actually is part of the report. It is already there.

DR. DOUGHERTY: But we really didn't discuss that. We really discussed are the 30 the right ones. We didn't discuss what the criteria should be in the future, unless you all did that at the first meeting which I did not attend. Do we agree with those criteria?

It could be part of our review of the report that we agreed to yesterday. But I think it is a larger issue than just reviewing the report. If the advisory committee is going to be making those recommendations, we need to identify some criteria in advance. I mean, you have to make exceptions to criteria and so forth and so on. It is not locked in stone, but I think we should get a better understanding among us of what we think are the important questions.

DR. HOWELL: Well, I think one of the things we can do in the future if it is deemed appropriate by the committee is to go back through the deliberations that are a part of that report and how those deliberations evolved, and what they included and so forth.

I do not think that should be a subcommittee effort. I think that sort of thing should come back to this committee if it is addressed. I agree with that.

Greg?

DR. HAWKINS: Just taking a kind of perspective on how we are doing this. If someone were starting a new lab someplace or starting a business to do this, what would they need to start this? I was sitting here breaking it down, and I think maybe we're trying to take everything and lump it too much together. But if we were say starting a company with doing all of this, the first thing we would want to have in place, and we've got it up here, is Laboratory and Standards Procedures.

Secondary would be probably Financing and Legal. Another area would be Education, and the fourth area would be Follow-Up. Also the thing we have to include in here is integrating all four of those areas, as IT is involved in all four of those areas.

So you have to consider if you were to throw IT off on its own, really IT is going to involve all these areas at one time. So you just can't kind of -- you've got to figure the structure there and how that is going to be involved if you set some of these things on their own.

Basically those were five key areas. I thought if someone were setting up a company, the things that they would have to deal with. Then the other thing would be how to prioritize them as far as how to set this whole system up if we were starting from the ground up.

Now, a lot of these things are in place, but that was just the way I was trying to think in order to prioritize it. The first thing I thought I would prioritize is you'd have to set up a laboratory, standards, and financing, and then you'd have the IT to start putting those things together. Then you start putting in the follow-up and the education.

But the problem is so many of these programs are already ongoing, you can't leave one component out. So some of these things we're trying to prioritize, and they really are all priority. How do you leave one of these parts out without it hurting the other? Especially IT. IT is part of all of these things. If you leave that out right now, you're going to hurt one of these other areas in integration later on.

DR. HOWELL: We have such a talented group around the table though, they can decide what the top priorities are, fortunately.

Coleen is going to start helping us there.

DR. BOYLE: No, I feel like my comments have already been said by others. I agree with those three top priority committees. I was thinking since we do have other pressing issues such as financing, evaluation, and IT, that each of those committees could think about some of those cross-cutting issues.

They won't be able to think of the whole picture, but they can at least come back and report on them, and then maybe in a second phase we can follow up in terms of the integration.

DR. HOWELL: That's an interesting idea, because the point is they do cut across all of the things that were discussed. I hear kind of an evolving feeling around the table for these top things, is that right? I heard some cross-cutting things about quality assurance from Denise. That means she'll have to serve on every committee to ensure that quality assurance is addressed.

But the other thing, IT is again, all these things require that to function. What is the sense of the group?

DR. HOWSE: I would certainly support what you are saying. Just to answer Denise's and Peter's question, at this point I would see financing as one of the cross-cutting issues. It may rise to a different level of importance as we continue to sort through. But I think for now, both the evaluation and the IT and the finance would probably best be taken on as cross-cutting issues across these three subcommittees, and then we'll see how it plays out.

DR. VAN DYCK: Can I make a motion?

DR. HOWELL: Please.

DR. LLOYD-PURYEAR: Can I just go back through what I have to make sure that you guys agree? You are going to change it?

DR. VAN DYCK: No, but -- okay. Go ahead.

DR. LLOYD-PURYEAR: Lab Standards and Procedures, Education and Training, and Follow-Up and Treatment. And as cross-cutting issues, what I've heard so far is evaluation and quality assurance.

DR. VAN DYCK: No, that's not quite it.

DR. LLOYD-PURYEAR: Wait. But I only heard once quality assurance. Information technology and financing. So not quality assurance, okay. I did hear it, but only once. Okay. That's all. I wanted to make sure I was --

DR. VAN DYCK: Can I?

DR. HOWELL: Peter has a motion.

DR. VAN DYCK: I'd like to make a motion that we have three subcommittees. To begin with, Laboratory Standards and Procedures, which includes new technology and the addition of new tests. Education and Training is the second, and Treatment and Follow-Up is the third, and that each consider the cross-cutting issues of evaluation, IT, and financing, and that we not put too many of these little bullets underneath yet, because I think that's up to the subcommittees to decide from the larger list, choose from the whole list, which elements they feel should be priority items for each of the subcommittees.

DR. HOWELL: Is there a second to Peter's motion?

PARTICIPANT: Second.

DR. HOWELL: We have a second. Further discussion?

Derek?

MR. ROBERTSON: Maybe what we could add as bullets at this point are the cross-cutting issues, since we have kind of decided on each one. So we just add like I think what Michele was just doing. You just have finance, evaluation, and IT under each.

DR. LLOYD-PURYEAR: Under each?

MR. ROBERTSON: Yes.

DR. HOWELL: Right.

DR. VAN DYCK: By themselves.

DR. LLOYD-PURYEAR: By themselves?

DR. VAN DYCK: Only. Just only those three.

MR. ROBERTSON: Right, right. Just only those three. And then like you said, Peter, the subcommittee would fill in the rest.

DR. VAN DYCK: I would modify the motion to include that suggestion.

DR. LLOYD-PURYEAR: Evaluation, IT, and finance.

DR. HOWELL: Right. But I think that the bullets that you have up there as we've been looking at it have been helpful, because you've seen what people are thinking about globally.

Joseph?

DR. TELFAIR: If that is what the committee is moving towards, I guess it is important, though, not to lose some of what has been discussed in terms of under these areas. Somehow or another if at least the notes can be maintained on that. I think everyone is going to have to at some point come back to those issues.

If it can be possible not to lose some of that other stuff, put it in some other places, notes, other areas to be considered, I think that would be important.

DR. HOWELL: I would tend not to destroy those notes.

DR. TELFAIR: Right.

DR. HOWELL: But I think not have them be directed to the groups.

Any further discussion? We've had a motion and a second about the three committees. Any further discussion?

(No response.)

DR. HOWELL: Can we see a hand of those persons approving this recommendation of three subcommittees?

(Show of hands.)

DR. HOWELL: And it is unanimous. Thank you very much.

MR. ROBERTSON: Mr. Chairman, I think you need to do all of these just before lunch.

DR. HOWELL: What?

MR. ROBERTSON: I said I think you need to do these votes just before lunch all the time.

DR. HOWELL: With regard to the fairly substantial discussions that we had yesterday about a variety of issues, I think those are very, very important discussions. I think that it is not only a consensus building technique, but also the committee is still very young in its developments, and it basically is kind of learning the territory and finding out what people think and so forth.

I think that it is sometimes an arduous pathway to democracy, which is very important and so forth. So I think that that was very important. Now, the final thing is that I want each of you, you can't go to lunch until each of you gives me the first subcommittee you would like to serve on, and an alternate two, which is all the other committees. Why don't you indicate the committee that you would really like to serve on, and then the committee of your second choice, provided that committee was completely filled with great people. That way, I will have all of those in.

The game plan would be to try to populate these committees, and then I will identify someone who appears to have great energies and great interest in that order, to chair the committee. You can communicate by email, by telephone, or other methods that you have, come up with an agenda, and I think that Bill's comment about trying to get those out electronically rather than to wait three months is a good idea so that we can see the agendas that the subcommittees have developed electronically, and this committee can say yes, that looks great, so you can proceed to work.

DR. BECKER: Yes, and also think about membership of the subcommittee. If, for example, you want a policy person from the Association of the State and Territorial Health Officers or something to interface with, or if you need someone from a clinical expertise field, or if you need to invite Brad, think about the membership of the committee once those committees are assigned. I think those are activities that can be going on while we are in the interval period of time.

DR. HOWELL: Yes. Clarify before we go to lunch, Peter, the mechanism by which persons not on this committee can serve as consultants, advisors, whatever, to these subcommittees.

DR. VAN DYCK: Well, with your approval, Rod, a subcommittee can ask for a presentation or consultation from basically anybody, which would include someone joining a phone call for answering specific questions or issues. I would think that would be the most used.

The next most used might be actual naming of somebody to one of the committees from outside. Again, they would have to request that of you, and you would have to approve that. So I think both of those mechanisms are possible.

DR. HOWELL: I will express my prejudices on that subject publicly before we go to lunch. That is that I think there is a tremendous amount of expertise in a variety of areas around the country, in the public sector and the private sector. I think that we should utilize that as and when appropriate.

(Whereupon, at 12:02 p.m., the meeting was recessed for lunch, to reconvene at 1:00 p.m.)

AFTERNOON SESSION (1:07 p.m.)

DR. HOWELL: Welcome to the afternoon session. We're already a bit late. We always appreciate the considerable public interest in the deliberations of this committee. Obviously the presentations become an official part of the proceedings, and as you know, are posted on the website. So I'm pleased that we have six persons who have signed up to comment this afternoon.

We'll begin with Rani Singh, who is a metabolic nutritionist, who will be our first person to present. Please go to the microphone, Rani, and hopefully it will come down to meet you there.

MS. SINGH: Mr. Chairman, thank you for giving me this opportunity to share my thoughts. As a genetic metabolic dietician, I want to open my comments by acknowledging that a majority of the diseases diagnosed by the newborn screening recommended by the ACMG panel, are treated by immediate nutrition interventions.

Therefore, the qualifications and the role of the metabolic dietician as a genetics team member can have a great impact on the outcome of these children. I wanted to emphasize the urgency for the need to train and support dieticians in the field. I was very excited to note the nutritionists clearly listed under "Direct Services" in Peter van Dyck's presentation yesterday.

This becomes even more critical in the face of shortage of biochemical geneticists while expanding newborn screening. I have created an international listserv for metabolic dieticians, and now we have close to 200 members. Participating members continuously express confusion with treatment protocols and lack of understanding with standards of care which lack

evidence-based research.

I feel well trained dieticians cannot only contribute to clinical practice, but also can contribute heavily in the area of research. I also want to thank HRSA for funding a pilot grant program in Region 3 for needs assessment for continuing education needs and development of the first educational module for nutrition professionals.

I do want to take the opportunity to urge that efforts to assure availability of qualified dieticians in the metabolic centers and the adequacy of low protein foods, medical foods are available for treatment. Lack of third party reimbursement continues to threaten the availability of treatment in some states to some affected persons, particularly those who have now entered adulthood.

We are in the process of forming a formal organization of metabolic dieticians to meet the needs, and feel confident that the group would be willing to assist the committee with any efforts focused in this direction.

DR. HOWELL: Thank you very much.

Are there any comments or questions of Ms. Singh?

(No response.)

DR. HOWELL: Thank you very much.

Our next person presenting is Ms. Judy Tuerck. She will be speaking on behalf of the Society for Inborn Errors of Metabolism.

MS. TUERCK: Editorial comment. I realized after reading this that I'm speaking to the choir.

Thank you, Mr. Chairman, for the opportunity to speak, and thank you to the committee for your efforts on behalf of those with inborn errors of metabolism and other genetic disorders. The Society for Inherited Metabolic Disorders, SIMD, is dedicated to improving scientific and public understanding about inborn errors of metabolism, and to promoting advances in the identification and care of those affected by inborn errors of metabolism.

Members of the SIMD are scientists, physicians, nutritionists, nurses, and other health professionals working in patient care and in research in the laboratory, in the clinic, in academia, in public health, in private medical systems, and in the biotechnical industry.

SIMD members provide diagnostic and treatment services to individuals of all ages with inherited metabolic disease to minimize the risks of disability and death. SIMD members play a prominent role in the diagnostic follow-up and treatment of children detected by newborn screening with inborn errors of metabolism. From this perspective, we very much appreciate your endorsement of the report "Newborn Screening: Toward a Uniform Screening Panel and System" at your last meeting.

We understand from presentations at your meeting that this endorsement is an important step along the path to expanding the panel of conditions for which newborns in the United States are screened, and to do so with uniformity.

We look forward to reviewing this report when it is available for public comment. In the meantime, we wish to take this opportunity to urge expeditious efforts to assure that newborn screening panels across the country are rapidly expanded to identify children with treatable inborn errors of metabolism. And two, that efforts to assure availability of adequate resources for successful newborn screening and follow-up.

The diagnosis if a biochemical genetic disease in and infant detected through newborn screening should be confirmed in a qualified diagnostic laboratory. Immediate and long-term treatment should be available from qualified and experienced experts in inborn errors metabolism. Mechanisms need to be in place to adequately fund all aspects of newborn screening, and to fund the treatment of inborn errors of metabolism in those who are identified by newborn screening.

Funding needs to be assured for education, testing, reporting of results, confirmation of abnormal screening results, diagnosis, long-term treatment, and evaluation of patients and outcome evaluation of newborn screening programs and practices.

Again, we thank you for this opportunity to speak and want to assure the committee that the SIMD and its members are eager to help you on behalf of the people we both serve.

DR. HOWELL: Thank you very much. I think that many folks were aware of the fact that this organization has among its members many of the people who are directly and specifically involved in treating the inherited metabolic diseases, many of most of which are identified with the expanded tandem mass spectroscopy.

We appreciate your comments, Judy.

The next person on the agenda that I have is Ms. Jill Fisch, who is a parent and is also the National Director of Education and Awareness of Save the Babies Through Screening Foundation.

Jill?

MS. FISCH: Thank you. Good afternoon. It is a pleasure to be here today, and an honor to have the opportunity to address the committee.

My name is Jill Fisch. I'm the National Director of Education and Awareness for the Save Babies Through Screening Foundation. I would like to acknowledge and say thank you to the committee and other agencies who are hard at work to move newborn screening in a forward and positive manner.

Collaboration between various agencies and organizations is essential. Newborn screening is not just an issue of the actual screening, it gives rises to other issues which need to be addressed. Some of the issues are education, financing, data collection, evaluations, and infrastructure. Other issues of great importance are linking identified children to services, ancillary services, manpower, money for treatments, and parental difficulties.

Some states are finally paying attention to newborn screening. Since the last meeting, Alabama, Connecticut, Louisiana, Michigan, Minnesota, New York, South Carolina, North Carolina, and Georgia have added disorders to their program. New York will be screening for all disorders by the spring, and Florida is going to begin adding disorders in February for a total of 25 added by the end of 2005. It is truly wonderful to see such progress being made.

However, there are states where there is no move to expand. It is my hope that upon Secretary Thompson's acceptance of this committee's recommendations that things will change. From those viewing this issue from a financial perspective versus quality of life, Colorado's newborn screening website states that "Identifying just two cases of PKU and 12 cases of hypothyroidism this year will save $2.5 million in lifetime costs for institutional care and special education."

Delaware's website states that "Newborn screening, together with rapid diagnosis and treatment, prevents mental retardation, illness, and death in newborns. It also saves millions of dollars in treatment, home, and institutional costs. For every $1 spent on newborn screening, $9 in medical care and treatment costs are saved, resulting in a national savings of $36 million every year."

Since the last meeting, approximately 530 unscreened babies with disorders have been born. I now view these babies as ticking time bombs. Time is of the essence.

There has been much discussion regarding the efficacy of treatment of certain disorders. One of these debated disorders, SCADD, has greatly affected my family. I have seen firsthand that treatment can be effective. My father has been receiving treatment for several months, and is finally able to manage the stairs in his home without difficulty.

We need to have a national database in place to track patients with these disorders and their response to treatment. This is the only way we will be able to see what treatments are effective. All states should be reporting their data to the National Newborn Screening and Genetics Resource Center so that needs are easily identified, and support can be given. I am not quite sure I understand why New York is the only state in the country that refuses to produce their data. This concerns me greatly. I am hopeful someone on the committee can help me understand why this is occurring.

I think education needs to be the number one priority as the subcommittees are developed and prioritized. We need to educate health care professionals about these disorders. The number of knowledgeable subspecialists is inadequate. My younger son had to wait six months to see a neurologist

who specializes in metabolic disorders. Raising awareness in the health care field will allow these professionals to choose subspecialties they had not been aware of before.

We also need to educate the health care professionals about the screening itself and the impact it has on families. One way to educate health care professionals about screening and the impact of these disorders is for professors to provide the education to their students. Families can be a great resource in the teaching process by coming to speak in classes. Hearing families speak would have a great impact, and would ensure that students actually remember the lessons.

Almost all professors should be able to arrange for families to come and speak. Many of these Professors are doctors who also treat patients, especially those affiliated with children's hospitals in major cities. Duplicate this effort nationwide, and you have education going on.

The other avenue for education regarding screening and disorders is free online CME. The Save Babies website gets 20,000 visits per month from professionals and parents all over the world. We have asked Pediatrix Screening to develop online CME in light of all the developments in newborn screening, and have assured them that we would be happy to put it on our website, or provide a link to CME on their site. This is now in process. We would be open to working with any other agency or organization interested in pursuing this avenue of education also.

Once the appropriate people work together and provide education to health care professionals, parents can become educated through their doctors. This is happening already in California. California is a shining example of effective parental notification, and should be followed by others.

The California Department of Health has circulated and continues to circulate the Save Babies pamphlet to the offices of every appropriate doctor and midwife with an insert specific to their state newborn screening program. We are receiving a steady stream of phone calls from the offices, and pamphlets are being ordered in record numbers. They are then distributed to pregnant women. We also have been getting inquiries from parents themselves who have been directed to us by their health care professionals, and they are all from California.

Lastly, I need to raise an issue greatly impacting the metabolic centers in New York. I am thrilled about the expansion of newborn screening in New York, however the program is being implemented without a fee. As a member of the Advisory Board of the metabolic center in the lower Hudson valley, I have been asked to help address this issue.

The metabolic centers in New York are unable to properly and adequately meet the needs of their patients without the fees generated from screening. There are no funds for repeat testing, follow-up, formula, and the other needs for families. I have reached out to the Governor's Office and the Department of Health to no avail. Our local center is now attempting to raise money through private donors to meet the needs of the center.

This is unfortunate, as the doctors at the center should be able to just focus on their patients and not have to deal with the aspect of raising funds. I am working with legislators in an attempt to rectify this situation and obtain the much needed funds for the centers. Any guidance that can be given to me by the committee regarding this matter would be greatly appreciated.

Thank you again for all of your hard work. I look forward to seeing these issues move forward at the direction of this committee and offer my assistance to the committee in any way it sees fit. We all have the same goal. We are all in this together.

Thank you once again for the opportunity to speak today. Thank you.

DR. HOWELL: Thank you very much for sharing your personal and other experiences with us, Ms. Fisch.

The next person on my schedule is Jana Monaco, who is a parent and also is on the Board of Directors of the Organic Acidemia Association.

MS. MONACO: Good afternoon, and thank you again for the opportunity to come and speak again on behalf of expanded newborn screening. As you recall, I have two children with isovaleric acidemia, one who was not diagnosed early and suffered severe brain damage, and one who was detected early and is living a very healthy, normal life, as any two-year-old should be.

I wish to thank each of you for the dedication that you have committed to newborn screening. We are all aware of the fact that it has not been an easy process, but you have diligently worked to finally get a report completed and prepared to send to the Secretary.

The mere fact that a report is coming has helped move things along in the State of Virginia, as well as other states. As pointed out yesterday, states are aware of the fact that they need to make changes, and Virginia is moving in that direction.

With all due respect, as the parent of a child severely disabled due to lack of comprehensive newborn screening, I have to disagree with Mr. Ross' statement yesterday stating that our newborn screening program is the high point in the American medical system. If it was, we would not be here today. It is good, but it is far from stellar, and it no doubt teaches us that we have a long way to go.

A recent example of this important fact is a baby named Joseph in Norfolk. He is a four-month-old baby diagnosed three and a half weeks ago with MMA, methylmalonic acidemia. His diagnosis came after weeks of diagnostic odysseys, only to be brushed off and attributed to the concerns of a young, single mother. Like Stephen, Joseph has been on a ventilator and the family has been told to consider turning off life support.

Over the past few months, I have spoken to the Virginia Genetics Advisory Committee at a local town hall meeting which has helped Virginia adopt bills in both the House and Senate to expand newborn screening to be consistent with the panel recommended here by the American College of Medical Genetics. The regulations must include follow-up and referral protocols and necessary provisions to implement the newborn screening program and any services available to the infants through the Children with Special Health Care Needs Program.

To help support the bill, I have been asked to speak on Monday to share my children's story before the General Assembly committee when the bills are read. I also had the privilege of speaking at the Northern Virginia Pediatric Society lecture in November, which was a great opportunity to educate physicians who are not up to speed with these disorders.

In all of my efforts, I have learned that people really do want to know about the subject and help bring about the necessary changes. It just isn't acceptable to continue to hear medical personnel admit that they do not know enough about inborn errors of metabolism. The emphasis should be on the education component in this process of expanding newborn screening, and it should be the number one priority in your subcommittee development.

Each time our children are hospitalized, we parents have educated yet one more medical staff person on these disorders. Recommending this universal panel is just the first step in a long process, and like me, other parents around the country are committed in their own states to help this process. Although I do not have your professional expertise, I do have more hands on experience with living with these disorders, like the rest of us parents.

This brings with it a parent perspective on the subject that is vital in the development of this newborn screening program. The concept of a database or communication system for health care for professionals is a critical component as well. There are already various models within the health sector to look at.

There seems to be such a concern for privacy in this whole process, yet I can assure you we lost our privacy the day Stephen was diagnosed. When he was first diagnosed, we had requested information about isovaleric acidemia. The physician tried her best to provide us with what she could, including some cases of IVA to read. Unfortunately, they were old cases off the Internet and did not provide us with much in the way of understanding the prospects of life with IVA.

It wasn't until we were connected with the Organic Acidemia Association that we learned a great deal about the disorder. That is where we discovered that Stephen was not the only one diagnosed beyond the newborn period, and that there were older IVA children and adults living with this disorder. This is where follow-up can truly educate.

We were introduced to families who had a great deal of information to share, information that we did not receive from the physicians. As you form your committees, seek to find ways to do long-term follow-up and possibly develop informational databases and use us parents in some sort of capacity. Our organizations are full of parents willing to help raise awareness, support research, and provide assistance with educating others.

They come with their own professional expertise and resources that complement our organizations. We are people that take initiative, in case you haven't already noticed that. I have already relinquished a level of privacy, like the others, for the sake of newborn screening and to connect with other parents.

My husband and I, along with three other IVA families, have developed a website called ivasupport.org whose goal is to be an outlet and resource website solely for families of already diagnosed and newly diagnosed IVA children. It contains stories and photos of children with IVA, a detailed definition of the disorder, newborn screening information articles, and studies on IVA by physicians, and a physician and dietician on board for questions.

We have provided a letter to our metabolic clinics and pediatricians to share with any new IVA families. As a parent, this is something that I wish had been available to us upon Stephen's diagnosis. When Caroline was diagnosed, it truly was a piece of cake for us. We knew what to do.

This is just an example of how resourceful parents can be. Again, I stress that you utilize us. We are a great resource for understanding the needs of raising children with inborn errors of metabolism and have much to contribute.

In closing, I speak for the Organic Acidemia Association in thanking you for moving forward to develop a universal newborn screening program that will reduce disparity among states and ensure quality and effective testing and treatment for babies and children in the United States.

Thank you.

DR. HOWELL: Thank you very much, Ms. Monaco, for those helpful comments.

The next person on my list is Dr. Phil Vaughn from the Pediatrix Medical Group. I think perhaps someone else will be speaking for Dr. Vaughn.

MR. SLIMAK: Yes. For those of you who know Dr. Vaughn, I am not Dr. Vaughn. Dr. Vaughn unfortunately has laryngitis, and his voice today ranges anywhere from Jack Webb to Harpo Marx. So I will make a valiant attempt to express not only the issues and concerns of the Pediatrix Medical Group, but also Dr. Vaughn.

Members of the committee and other attendees, thank you for this opportunity to address you. My name is Bill Slimak, and I am Vice President of Operations for Pediatrix Screening.

Just a brief description of my background. As you know, Dr. Vaughn is a neonatologist. My background, I am formally trained as a chemist, specifically a clinical chemist with 30 years of experience, and I know that's hard to believe, but 30 years of experience in health care and pharmaceutical operations, specifically in licensed biologics. So I come with not only manufacturing experience and I have worked for companies like Johnson and Johnson and Bayer, but also high volume clinical lab experience.

For five years I was General Manager of Laboratory Operations for the New York Blood Center. So I come with experience in not only operations, but operations in a highly regulated industry, which would talk to the concentration on quality and continuous improvement.

We are proud to be part of this enterprise as we move forward. Pediatrix Medical Group's mission is to improve the lives of newborns. Screening is an important part of that mission. Pediatrix Medical Group has been a leader in not only newborn hearing screening, but now in newborn metabolic screening. Specifically, Pediatrix Screening was instrumental in developing the application of tandem mass spec to newborn screening, and now has some ten years and 2 million babies experience operationally, technically, and clinically.

This is not only from the screening standpoint, but we also have unfortunately substantial experience in background and postmortem screening, which is unfortunately a part of making sure a feedback mechanism, to making sure that cutoffs and standards are kept in check.

We are pleased that the ACMG report is now moving forward and will be made available to the Secretary and to the public. This is an important step in encouraging the adoption of broader scope of screening services, and we obviously support that.

Pediatrix Screening and Pediatrix Medical Group anticipates that through our participation in upcoming subcommittees, we will be able to substantially contribute given our experience and depth and breadth of expertise. Just to go through some of those. In screening technologies, as has been discussed here, we have a lot of experience not only in primary screening, but what we call second tier testing. We have broad experience in not only biochemical techniques, tandem mass spectrometry, and then also molecular genetics, which a lot of the second tier testing involves that technology.

Research and education, Pediatrix Medical Group, in the arena of neonatology and pediatrics is recognized as an entity that brings forth not only research, clinical research, but also an educational piece. Data management. On an annual basis, we look at some 250,000 deliveries and patient days nearing one million patient days. We have this all in a database, and we use this database to help us do best practices, reevaluate clinical situations, and put forth in those the best practices that can come out of that.

Clinical trials, because of our vast involvement in pediatrics and neonatology, we are often involved in the clinical trials of new and interesting not only medications, but techniques and procedures. Standards. We also recognize the importance of standardization where possible. Pediatrix Screening actively participates in important standardization committees to ensure patient safety, and build continued confidence and capability as it advances.

One of our scientists is in fact the chairman of a NCCLS committee. Anyone who is involved in clinical laboratories understands that that is a group that drives consensus and puts out guidelines for standardization. He is the chair of that committee, and they are specifically looking at tandem mass spec with newborn screening as a part of that standard setting, and obviously operational controls. We deal in a high volume situation, in a situation where the reality is not too long ago, tandem mass spec sat in a research laboratory, and the transition from a research laboratory to a production or high volume laboratory, you talk about implementation, that is one of the biggest challenges, to take that device and turn it into a production device.

In closing, we have the operational, technical, and clinical expertise to help this committee in reaching our mutual goal, ensuring that newborn screening is available to all in a very comprehensive and quality manner. These issues are complex, and states, parents, and providers will be looking to all of us in the newborn screening community to help.

To omit Pediatrix Screening from the process I believe does not further the goals or mandates of this committee. For these reasons, we respectfully request the opportunity to participate in a direct and positive way.

DR. HOWELL: Thank you very much, Mr. Slimak.

I'm pleased to introduce at this time Ms. Micki Gartzke, who is a parent and also is representing the Hunter's Hope Foundation.

MS. GARTZKE: Good afternoon, Mr. Chairman, and members of the committee. Thank you to you and the many collaborating agencies and partners for your work on behalf of the children and families in the United States. This joint effort is invaluable to American families. It is truly my pleasure and an honor to be able to share comments with you today.

As Dr. Howell just shared, my name is Micki Gartzke, and I lost a 2-year-old daughter to lack of early identification of Krabbe disease. I am now the Director of Education and Awareness for the Hunter's Hope Foundation. We at Hunter's Hope are committed to all children having equal access to all available newborn screening.

Along with the many thousands of families that we work with who have now gained awareness of the need to improve newborn screening on a national basis, we applaud you all for your hard work and your commitment towards fixing this public health program. We are very excited to hear about the progress that will come soon with the forward movement of the ACMG Report. With more and more involvement and greater partnership on the variety of issues still to be solved, more children's lives will be saved, and that is a great asset for America.

Resolving of the agenda items will bring faster newborn screening and consequent follow-up care to children. That is what is at stake, providing effective treatment to those children identified through newborn screening. We think the priorities for the subcommittees should be driven first and foremost by the children's needs. Right now, that is education.

As we expand data collection nationally, we will find out what is working best so that we can apply it broadly across all the states to help health care professionals do their jobs better. With expanded funding and funding strategies, equatability will become a reality, and we will realize more effective treatments.

With all states reporting their data to the collective resource of the National Newborn Screening and Genetics Resource Center, those states with needs will be further identified, and support can flow where it is needed. With expanded infrastructure, opportunities will be more broadly available, linking the children to the necessary services that are of the utmost importance in this process.

I continue to hear of children having six-month waits to see pediatric neurologists. I continue to hear of medical centers not having adequate resources to give the proper care. These issues need to be fixed so that children can receive the best care possible. We need to get real newborn screening educational information into the hands of expectant parents so that they can ensure that their newborn is receiving the best possible option for newborn screening.

We need to engage the obstetric providers to provide this information. We need to educate our medical students about newborn screening. With education available at the medical school level, the importance of the entire newborn screening process will generate more physicians filling the service gaps that exist today. This education can be strongly supported by partnership, including parents who can provide real life experiences.

We have seen that the states are listening and have made some progress. Thank you, Dr. Therrell, if you are still here, for your wonderful presentation yesterday. I was just really pleased to hear all of that.

Thank you also Jill Fisch, because you highlighted some of this information in your comments earlier.

Just this morning I received on email some new bills in Kentucky. So they have new newborn screening bills moving forwarding Kentucky, and I think that's a pretty good sign considering it is Kentucky. But still many of the states are lagging, not only in the area of providing the adequate newborn screening, but even in getting the word out in an effective and meaningful way to parents.

We heard today that California has made a real effort to get this vital information into the hands of the relevant physicians and midwives, and these health care professionals are then getting the word to the parents in a timely fashion. This is helping them to raise their standard of care in newborn screening.

It seems to me and other advocates there are still many questions that need to be answered, and I know you all will work hard to find the answers to improve these newborn screening standards nationwide. A couple of questions that come to the top of my mind, where are the parents still having difficulties? Are we spending enough money on treatment? Are there adequate subspecialists geographically located to provide the necessary services? What can be done to motivate the still resistant states to expand their services? We will do whatever we can to help, and we are looking for your guidance on how we may expand our advocacy efforts.

Hunter's Hope started in 1997 as a foundation with the mission to increase public awareness of Krabbe disease and other leukodystrophies, to increase the likelihood of early detection and treatment. Of course we now know that access to lifesaving treatment lies in the hands of expanded universal newborn screening.

We started out to save the lives of children born with Krabbe and related leukodystrophies, and now through our strategic planning, our fight for early identification has expanded to all diseases that have newborn screening tests to help save children's lives in America.

To date, in addition to providing more than $4 million towards leukodystrophy research, Hunter's Hope has developed a multifaceted universal newborn screening campaign. Some of its recent activities are listed in my written comments. Notably, we have reached out and met with state officials in New York alongside Ms. Fisch, working to get New York State to expand its program.

We are in the process of supporting a pilot program in New York State for newborn screening for five lysosomal storage disorders. We are advocating getting additional states in this pilot program. Wisconsin has indicated a preliminary interest in participating. Next month I will be in North

Carolina seeking its participation. We are providing blood spots of affected Krabbe children and healthy siblings for this pilot. We are reaching out to other lay advocacy organizations to expand this support.

We plan to meet with various transplant doctors from around the country as we host our 8th annual medical and family symposium this July in western New York to expand the treatments for newborns with leukodystrophies and related lysosomal storage disorders.

I thank you again for your time and your hard work. We all know that the children can be saved, and that's our mutual goal. We know that you are committed to make that happen as quickly and effectively as possible, and we want you to know we'll do whatever we can to help. We hope that you will let us help.

Thank you.

DR. HOWELL: Thank you very much, Ms. Gartzke, for those comments.

We now are going to hear from Joyce Hooker from Colorado.

MS. HOOKER: I'd like to thank the Genetics Branch and Hereditary Diseases Program for funding the Regional Collaborative Centers. I have been the Regional Coordinator for the Mountain States Genetics Network since 1985. It almost makes me older than dirt.

Our network includes Arizona, New Mexico, Texas, Wyoming, Utah, Colorado and Montana. We go from Canada all the way down to Mexico, so we're quite a large area.

Over the years, the regional networks have produced numerous pamphlets, brochures, booklets, professional journals, articles, collected data on newborn screening and clinical services, conducted surveys, prepared reports and physician papers, created legislation, and set up clearing houses for materials, just to name a few items.

But the most important and productive activity has always been the opportunity to network. This is the backbone of the Mountain States Genetics Network, now the Region 6 Genetics Collaborative Center. Networking is the first step to collaboration and coordination of services. It can take on many patterns and designs.

There is what we refer to as both informal and formal networking. Formal networking being a designated time and place, such as we have today, or informal, whenever it happens. It can be at the swimming pool, it can be at the bar, it can be on the street, it can be anywhere. It can take place on the phone, the Internet, or via the many telecommunication systems available.

But the most effective and valuable means of networking is the age old face to face meeting. Each year the network asks it members in an annual meeting which activity or facet is the most valuable to them. It is always the mid year committee meetings and annual meeting.

The network has seven multidisciplinary committees, each on a particular issue. Whether it is the Consumer Issues Committee or the Laboratory Practice Committee, it is comprised of consumers, physicians, genetic counselors, laboratory staff. The network is committed to providing each committee a full day face to face meeting in the winter, and a three-hour segment at the annual meeting in July.

This supports the committee member's time and opportunity to meet and know their peers and others in the region to sort out and discuss issues and problems, and to plan strategies and activities the committee will carry out and share data and ideas in a comfortable, non-confrontational

environment. This doesn't just happen on its own. It takes commitment and work from those attending and the staff.

But with the benefits in this great age of electronic technology and communication, we cannot forget the human element. After all, that is what we are all about.

Thank you for your generous support to the Regional Collaborative Centers. We will serve you well.

DR. HOWELL: Thank you very much.

That is all the persons who have filed to present today. We appreciate all that public comment. So I'm pleased to announce that we're a little bit ahead of schedule, which is always a good thing.

DR. BECKER: Rod, there's a hand in the back. I don't know if you want to acknowledge that or not.

DR. HOWELL: Oh, I'm sorry. Apparently one person got left off my list, for which I apologize. That is Dr. Sybinsky from the Association of Maternal and Child Health Programs.

DR. SYBINSKY: Thank you very much, Mr. Chair, and members. I'm Peter Sybinsky, CEO of the Association of Maternal and Child Health Programs.

AMCHP represents state and territorial public health leaders who administer statewide programs to improve maternal and child health in many, many different ways, from newborn screening systems to clinics for children with special health care needs, school-based health centers, and a wide variety of other vital health programs.

We'd like to thank the committee for your attention to this critical public issue, and today we would like to speak to you briefly on two aspects of that issue. The first is the importance of addressing the entire newborn screening system, including follow-up and the key role of state family health programs. Also the second dimension of adequate resources to support these state newborn screening systems.

Testing newborns for metabolic disorders is essential to giving every child a strong start in life. As you know, and as Dr. van Dyck outlined yesterday, newborn screening is more than just testing. It is an entire system that includes parent and doctor education, medical referrals and follow-up, ongoing financial support for a child's medical care, tracking and quality assurance, among other processes.

Maternal and child health agencies across the nation help establish and direct newborn screening systems in every state. These are complex systems which go well beyond screening. Their objective is to ensure that families don't fall through the cracks. States do this in different ways, but with the same thing in mind. Assuring that a positive screening test results in some closure, whether it be treatment or assurance that the condition does not exist.

As the committee moves forward with its work, AMCHP urges you to consider all aspects of newborn screening systems. Not only the actual blood testing, but also the extensive follow-up activities and oversight by the health providers and the state maternal and child health programs. The timeliness and accuracy of testing and follow-up services can make a tremendous difference in the life of a newborn.

However, as we know from Kay Johnson's presentation at your previous meeting, these activities come with a price of over $120 million the GAO says was spent on newborn screening in 2001, only 26

percent paid for follow-up services. While states rely significantly on the fees charged to families or hospitals to finance testing and laboratory costs, the Maternal and Child Health Block Grant usually pays for follow-up services and specialty care when it is not available through Medicaid, CHIP, private insurance, or other sources of funding.

As newborn screening programs expand, someone is going to have to pay for this additional testing and follow-up. States continue to face budget limitations and expanding public health responsibilities. In this environment, it is difficult for states to maintain existing services, much less meet the needs of an expanded screening program. It is also unrealistic to expect families to pick up the additional cost of screening.

In this environment, federal support for programs like the MCH Block Grant and Medicaid is therefore critical. AMCHP encourages the committee to recommend that the Secretary support increased funding for the Maternal and Child Health Block Grant in the President's budget so that our state Maternal and Child Health Programs can meet the increased needs of expanded newborn screening programs.

AMCHP also calls on the committee to recommend full funding of the Title XXVI of the Children's Health Act of 2000 and increased funding for the efforts to help state newborn screening programs develop integrated data and surveillance systems so important to linking families to follow-up and treatment.

In addition, adequate funding for Medicaid and CHIP will be vital to the success of expanded screening programs. If a child can't get expanded medical services because they can't afford them, all the value of increased screening will be lost.

In conclusion, AMCHP strongly encourages this committee to consider the full cost of newborn screening programs, including effective follow-up activities, and recommend adequate funding to link all families with appropriate care. The recommendations of this committee can greatly strengthen state newborn screening systems and maximize the value of the additional screening tests.

AMCHP thanks you for your efforts and looks forward to providing any assistance we can as you move forward in your work. Thank you for the opportunity to provide input to this important public health effort.

DR. HOWELL: Thank you very much, Dr. Sybinsky, for your presentation and your written material. We apologize for having you off of my list.

Have I missed anyone else that has prepared material that we don't have?

(No response.)

DR. HOWELL: Excellent. Thank you very much. Again, we're still ahead of schedule. Let's move ahead if we might into the committee business situation.

Before lunch, we had a productive discussion of subcommittees and so forth. Each of you provided notes which we dutifully collected before lunch about your first, second, and final choices and so forth. There was fortunately a really broad interest, so that there was a scattering and so forth, and so we were able to formulate some subcommittees at lunch that accommodated most people's first, and in all cases, at least your second choice of work.

One of the things as you look at the committees, however, is that people who ended up in the Education Subcommittee also has expertise in the other areas. Obviously we would hope that as these

efforts go forth, you would draw on other members who have a great deal of strength and activity in that area.

The committees came out in the following way. That is under Education and Training, the persons who signed up for that as either their first or second things were Drs. Howse, Edwards, Becker and Hawkins. I would like that to be the members that serve on that subcommittee. I asked Jennifer if she would be willing to chair that subcommittee, and she has generously agreed to do that.

Under the Follow-Up and Treatment Subcommittee, we had Drs. van Dyck, Dougherty, Boyle, Robertson, and Telfair who listed that. That is obviously an important committee. That's the only one that has five members. I asked Coleen if she would be willing to lead that group. Denise, in spite of the fact that I threatened to put you on all of the committees, that's the only one I put you on. But we expect you to talk about follow-up issues on all of these.

Under the Laboratory Standards Subcommittee, that was an interest of virtually everybody, not surprisingly. Of many people. Not everybody, but many people. The persons that we put on this list are Drs. Alexander, Coggins, Brower and Rinaldo. I have asked Dr. Brower if she'd be willing to serve as chairperson of that committee, and she has been generous in agreeing to do that.

So that's the way the list came out and so forth. I wonder if there are any comments about that from any of the committees.

(No response.)

DR. HOWELL: I think it was an excellent assortment of talent and so forth. Now, as I understand what we are going to do now is that the folks that I have asked to chair these subcommittees will communicate with the persons that were listed on your committees and have some preliminary discussions, planning, and so forth. Then develop some plans of actions, some agendas, whatever you want to call it. But basically what you hope to do. We can then circulate those. Michele can circulate those through the whole committee and keep records on those, and we can look at those and so forth.

Prior to the next meeting, there is no reason that substantial activities can't come forth as far as some work products or certainly some definitions of what you plan to do. Again, the protocol is that if there are people that you would like to involve on the committees and so forth as you proceed after you have the discussions with your group, if you want to add those people, you'll need to touch base with me since that needs to be done officially through the chair of the committee. I'll be glad to work with you.

Is everybody on the same page? Is that what we are planning to do? Again, as you look at these little groups, and they are very small groups there, in virtually every instance there is a person that has a very strong interest in one area that the committee would be looking at. Some might have interest in current lab cutoffs and so forth, the technical aspects of the current tandem mass spectroscopy such as Piero. Some of the other groups would be interested in technology developments. So there is an opportunity for a spread there and so forth.

Peter?

DR. VAN DYCK: I may have been daydreaming for a second. But did you set a time when our agendas should be in to you for approval? Some interim time before the next meeting.

DR. HOWELL: I did not set a time, but you were very courteous in suggesting that I think of a time.

(Laughter.)

DR. HOWELL: That's why you've been so successful in politics.

(Laughter.)

DR. HOWELL: But the thing is is what time frame should we talk about?

DR. BOYLE: It might be helpful too to know exactly what we need to have in by this time. It is the agenda as well as the members, any additional members that we might want to have as part of that committee, is that correct?

DR. HOWELL: Yes, and I would suggest that we start off without recommending additional members, but talk about maybe people that would serve as consultants to you as opposed to formal members at this point. But I would suggest that we consider formal members as time matures a bit. But I think that's correct. What is the time frame that would be logical? A month? Is that too soon? It certainly doesn't seem too soon. The next meeting is April. We have a vote to my left for the end of February. That's a bit more than a month. I see noddings of heads. So we'll expect those to be back to us by the end of February then.

Bill?

DR. BECKER: Rod, as we obviously are going to become more active in the intervening periods now, was there an attempt, and maybe I missed this because I get a million emails, to set up a listserv for the committee so that we could share things amongst ourselves? I thought that came up at one of the previous meetings.

Well, let me ask the question then. Michele is not remembering it. Is it possible to set up a listserv for the Secretary's Advisory Committee? That would be one vehicle to distribute information to the group.

DR. HOWELL: There is a conference over here on the subject.

DR. LLOYD-PURYEAR: I prefer, and in fact, I am requesting that it be done through me, just because of sort of keeping control of committee business to make sure that staff at the bureau, since we are in charge of staffing, are aware of what is going on. But that's easy to do.

DR. BECKER: That makes perfect sense to me.

DR. LLOYD-PURYEAR: And it is also security.

DR. BECKER: I understand, I understand. So in the interim time, any information that we want to distribute to the committee, send it to you?

DR. LLOYD-PURYEAR: Yes.

DR. BECKER: That's fine. I mean, it is one step removed, but it works.

DR. HOWELL: Okay. Any other issues on the committees and so forth? So I think that by the end of February, we will anticipate some information.

Anything else we need to do on those, Peter?

DR. VAN DYCK: Just so we can be straight, the task of the committees is to come up with an agenda that gets approved by you before the next meeting. Is there anything we should do then if you turn around those approvals say in a week or 10 days or something after the end of February, is there an expectation that we do a little more than that before the next meeting? I wonder if there is going to be a discussion among the chairs or among us here. Should we wait until the next meeting and come prepared to really work on the agenda at that point?

DR. HOWELL: Thoughts of the committee?

DR. BECKER: Rod?

DR. HOWELL: Yes?

DR. BECKER: Maybe I can offer a suggestion. Maybe not what we -- well, in addition to an agenda, perhaps an attempt at prioritization or an action plan. Really I was thinking of an action plan as to what activities the subcommittee feel are most important and would, once receiving approval, would start moving in those directions. That is the kind of information you would want to see at the six-week mark.

I think, Peter, that would answer your question of what would happen next.

DR. VAN DYCK: I guess what I was going towards is if that could be done, then perhaps what the subcommittees have as their priorities might drive the agenda, the meeting, a little bit in relation to what speakers came or whatever.

So we probably could if the approval for the agenda or action plan could come by the first week in March or something --

DR. HOWELL: That would not be a problem.

DR. VAN DYCK: Then that probably would be soon enough to help drive which speakers came for your choice, Rodney. I don't know what kind of speakers came and who came.

DR. HOWELL: We could do that.

Joseph, you had a comment.

DR. TELFAIR: My question has been answered.

DR. HOWELL: Any other questions? The other thing that we need to talk a little bit about is the future agenda. We've talked about these issues and so forth, but are there other issues that we should have on the agenda for the next meeting that someone has a burning interest in as far as people that we know, regardless of what comes out of these workgroups or subcommittee groups that would be important to have presented here?

DR. BOYLE: I guess I might suggest, and I had to think this one through a little bit more. But those cross-cutting issues. I mean, since they will be relevant to all of us, Education, IT, the Financing, what was the other one? Evaluation. Perhaps we could flush those out a little bit more as a group in thinking through.

DR. HOWELL: Okay. And that would entail getting some persons to come and address those that we can have as an issue there. Michele can start working and thinking about that and communicating with you.

DR. LLOYD-PURYEAR: You wanted all three areas? Or just Evaluation?

DR. BOYLE: I was thinking all three.

DR. HOWELL: Any other issues here?

Bill?

DR. BECKER: Rod, I would think a continuing item for our agenda for at least the next meeting, and maybe beyond, is a continuing status report on the ACMG product. I guess it will end up being the HRSA report as produced by ACMG by that point. Hopefully if all goes according to plan, that report will have been out in the public arena for some time, maybe still in the open public time period, but it will also be an opportunity for if it has been officially released by the next meeting of this group, it is obviously a document that we have not seen all the final product of and will need to probably have some time to discuss or review some of the areas that we've not formerly seen, like the ACT sheets and some of the other things.

DR. HOWELL: It would be highly likely, I would certainly hope, that this document would be in the public arena, and members of this committee will have had the document some time. So it would be appropriate to have discussions of the document.

DR. BECKER: I would add a companion request to that. It is basically the same vein. We have asked Mike to come and give a quite extensive series of two presentations on of course the uniform condition panel, but he has, because of time and necessity, not talked a lot about some of the other program standards portions of this document. I think those are germane to what the subcommittees are going to be working on, and perhaps to try to get a presentation on those aspects of the report that we haven't already heard about.

DR. RINALDO: I have a question for Peter and Michele. Because I think although we have, well, I know what we have, or at least mixed feelings about how we have a process for the report. But I actually would like to know what happens next beyond this, what is going to happen to it. Once it goes to the Secretary, what the options are. So I don't know who is the right person to educate us, but I think we should also continue to follow the path of that report and the recommendation of the report.

DR. VAN DYCK: Well, we can certainly report on the status at each meeting. I'd be happy to do that.

DR. HOWELL: It would seem to me that it would be important to have a status report, and then again, reports that we've discussed here.

Coleen?

DR. BOYLE: Well, I thought we voted yesterday on coming up with some sort of collective thoughts from this committee about the final report.

DR. HOWELL: Yes.

DR. BOYLE: I don't know how we're going to do that, so we might want to just talk about that process.

DR. HOWELL: I would think that one part of that process would certainly be an opportunity to look at it in some detail, as Bill has been talking about.

But what thoughts do you have, Greg?

DR. HAWKINS: Well, and one other thing we talked about is I don't know what timing will be appropriate for the next meeting, but information that we get back, public comment on the report. I don't know, if it goes out, will that be back in time for us for the next meeting to discuss?

DR. HOWELL: I think one of the questions is that the time limit has not yet been established for public comment.

DR. HAWKINS: Right.

DR. HOWELL: That will be decided in conjunction with the department. So I would think if it is a 30-day period and we are granted access to the public comment which we would request, I guess we could have some back then. But also we clearly could not.

DR. RINALDO: Greg, you said something about your plan when you start up something. You mentioned the legal aspects. Should we pay some attention to the legal aspects related to newborn screening discrepancies? Is that a topic we would like to hear about what is going on?

DR. EDWARDS: Well, I think there are other things involved in legal aspects other than the testing, too. The legal aspects of the way the system functions, who falls through the cracks. I think we should look at all of the legal aspects of the entire program, not just of the testing.

DR. RINALDO: Well, I wasn't thinking about the legal aspects of the testing, but I think it is somewhat becoming a reality that it is in part both a driver and driven by what we are doing here. You cannot deny that there is legal action out there related to the provision of screening. Is that something we want to discuss?

DR. EDWARDS: I think we want to discuss all of the -- you are saying the legal aspects of screening versus not screening. I think the legal aspects, which I thought I heard you say earlier, I think we should look at all of the legal ramifications of the program.

DR. RINALDO: Well, I wasn't in any way limiting. Again, it is just an idea there. Is that something you want to have somebody to present and review?

DR. HOWELL: Denise, do you want to comment on that?

DR. DOUGHERTY: Yes. That sounds like a very important thing to talk about. I'm just wondering if we want to not do it the meeting after next, because if we're discussing the agendas for each of the subcommittees and we're getting presentations on each of the cross-cutting issues and discussing comments on the report, that might be a full agenda. That will be a full agenda.

DR. HOWELL: I gather that there is interest, however, in having a discussion about the legal ramifications of the newborn screening program. So maybe we can end -- well, yes, the ELSI issues as well as the others. That is something that maybe we can have Michele put on the agenda. Next time is clearly going to be really busy. We do have legal members of the committee, but it would be helpful perhaps to have some outside folks that focus in this area to come and address some of the legal issues. That's an interest in the medical practicing community obviously, and there are obviously ethical issues that certain people we could get to come.

DR. HAWKINS: I was just going to make a suggestion that we have all the three subcommittees. As we meet and start discussing the legal issues that pop to mind, we should jot them down and give them to you for the next one. I think that would give us a good idea of where to go.

DR. HOWELL: That's a very good idea. A lot of that will come out of Coleen's group I think. Anything else?

DR. TELFAIR: Yes, a couple of things. First of all, I'd like to recommend a name to talk about ELSI issues, which would be Vince Bonham from the Human Genome Project. That is what he does, and he would cover this issue fairly well. I recommend him for future reference on this.

DR. HOWELL: Thank you.

DR. TELFAIR: As well as I think he would cover it pretty thoroughly. The other thing is I think if you are putting things on a future agenda, you have evaluation on the agenda, but also the other aspects of utilization of both the evaluation and the information. Some discussion from the point of view of those who are utilizers, from representatives of different groups, consumers, providers, you know, data persons. As you did with data, you have different groups representing it, I think there may be education to this group, to this committee, to have different perspectives on that as well. Not like right away, but as you are putting things on a future agenda, I would recommend that.

DR. HOWELL: Thank you very much.

Denise?

DR. BOYLE: We didn't hear who you recommended. I'm sorry.

DR. TELFAIR: Vince Bonham.

DR. DOUGHERTY: I guess you're recommending people. What is the best way to, if we know of people on these various cross-cutting issues, to get ideas to you for speakers?

DR. HOWELL: Let me know. I develop the agenda with Michele.

DR. DOUGHERTY: Okay.

DR. HOWELL: We fortunately have an embarrassment of riches. So there are a lot of very good people that we can call on who have focused in these areas that can really be helpful to us as we think about it. But if you could let me and Michele know. Michele has a better memory.

Anything else that we need to discuss on that particular issue? Piero?

DR. RINALDO: There's another word that hasn't been mentioned much, at least at this time. That is research. So I know it was one of the topics in the list, so that's another one for later.

DR. LLOYD-PURYEAR: But isn't that captured somewhat under test development?

DR. BECKER: And it's listed in Peter's algorithm. I think all of the subcommittees are charged with taking a look at that list and applying it as fits their particular areas.

DR. HOWELL: But I think obviously research will be an important thing. I don't think there is much of a chance that will get forgotten, but it will certainly appear on the list.

I would think that as the groups work toward some of the immediate solutions, one of the things that will come out is areas where the knowledge is incomplete. That will obviously suggest a great

research agenda. There are obviously research agendas well underway to look at some key research areas surrounding newborn screening. That is in Dr. Alexander's court at this point in time.

Any other things on the agenda? The agenda, goodness, we're going to have a four-day meeting next time. But anyway, the future agenda. And as always, while all the public is here, we want to be sure that we always allocate time for public comment. It is important that we know in advance that you're coming. It is also very nice when you have a prepared thing to put in the record, because that is very accurate. So we appreciate that very much.

The other thing, before the evening slips away, that is that we are currently scheduled to meet in if you look in the very last thing in your book, we are scheduled to meet in April on the 21st and 22nd of April. That is scheduled as a tentative date. I'd like to have you look at that to be sure that that is the best date that we can find.

Michele has worked diligently by email and so forth to look at that. We are scheduled for the next three meetings, the next tentative meeting is on July 21st and 22nd, and October 20th and 21st, all in this room, which I personally think is a very good room. It gives us more space, and it is elegant sitting atop this building. It lends credence to our efforts, which is important.

I told someone who was concerned about this next schedule, if we had to change the schedule and meet in the terrible hotel again, we were going to say we were in that dreadful hotel rather than in this dome cathedral because of them. Would anyone like to comment on the dates?

DR. EDWARDS: I think the main thing is just knowing about it ahead of time is so important. I would hope at the next meeting, we could then pick up the January meeting for next year.

DR. HOWELL: Okay.

DR. EDWARDS: I mean, by knowing this far in advance, at least it helps many of us as far as setting our calendars, and we can do that.

DR. HOWELL: I think it's very helpful for everybody. I think many people have a problem with making commitments so far in advance. But I think that setting the dates as far in advance as possible will help more people get here.

When this meeting was originally tentatively scheduled, we had it scheduled for the same day as the inauguration. So things like that come up, but we want to be sure that we're not doing anything like that. Can you then put those dates in your calendar, and we will consider those firm unless we hear something. Is that reasonable to put them as the best dates at the current time?

DR. TELFAIR: I agree with Stephen. Just knowing in advance, particularly a couple of months in advance, is always an advantage.

DR. HOWELL: Okay. Good.

I wonder if there are any other things that we need to discuss before we leave. We are ahead of schedule, which is always a good thing, and which I always like. But unless there is some compelling issue that is going to change the face of the world -- Derek, I don't think you were here, but you are on the Follow-Up and Treatment Committee, which is very carefully and credibly headed by Dr. Boyle, who will keep you working hard. She will be in contact with you soon.

MR. ROBERTSON: All right. Thanks. I thought I was on the Lab Standards and Procedures.

DR. LLOYD-PURYEAR: I was joking.

(Laughter.)

DR. HOWELL: That was the only one you didn't choose, so we put you on that one. No.

MR. ROBERTSON: I apologize for not being here. Something came up at work.

DR. HOWELL: Can we have a motion to adjourn the meeting?

PARTICIPANT: So moved.

DR. HOWELL: And second?

PARTICIPANT: Second.

DR. HOWELL: We like to be formal. Thank you very much for your attention. Have a safe journey home.

(Whereupon, at 2:20 p.m., the meeting was adjourned.)